

GenCore version 5.1.9
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OM protein - protein search, using sw model

Run on: September 11, 2006, 22:15:38 ; Search time 209.448 Seconds
(without alignments)
4398.662 Million cell updates/sec

Title: US-10-632-342-8
Perfect score: 10489
Sequence: 1 MANFLPRGTSSFRFRFTRES.....HSEDLADPPSPDRDRESIV 2015

Scoring table: BLOSUM62
Gapop 10.0 , Gapept 0.5

Searched: 2589679 seqs, 457216429 residues

Total number of hits satisfying chosen parameters: 2589679

Minimum DB seq length: 0
Maximum DB seq length: 2000000000

Post-processing: Minimum Match 0%
Maximum Match 10%
Listing first 200 summaries

Database :

A Genesep2_8:.*
1: Genesep2_1980s:.*
2: Genesep2_1990s:.*
3: Genesep2_2000s:.*
4: Genesep2_2001s:.*
5: Genesep2_2002s:.*
6: Genesep2_2003as:.*
7: Genesep2_2003bs:.*
8: Genesep2_2004s:.*
9: Genesep2_2005s:.*
10: Genesep2_2006s:.*

Pred. No. is the number of results predicted by chance to have a score greater than or equal to the score of the result being printed, and is derived by analysis of the total score distribution.

SUMMARIES

Result No.	Score	Query Match	Length	DB ID	Description
1	10489	100.0	2015	8	ADM34001 Human SCN
2	10487	99.9	2015	7	ADF56441 Human Nav
3	10484	99.9	2015	8	ADM33999 Human SCN
4	10484	99.9	2015	8	AE822972 Sodium ch
5	10478.5	99.9	2016	8	ADM33997 Human SCN
6	10478	99.9	2015	8	AE822993 Mutant hu
7	10478	99.9	2015	8	AE822994 Mutant hu
8	10473.5	99.9	2016	8	ADM33995 Human SCN
9	10473.5	99.9	2016	10	AEF90518 Human SCN
10	10471	99.8	2015	9	AE78664 Human SCN
11	10432.5	99.5	2016	4	AA882239 Human SCN
12	10432.5	99.5	2016	7	ADDE44756 Human Pro
13	10432.5	99.5	2016	7	ADDE55106 Human Pro
14	10429.5	99.4	2016	4	AA882245 Human SCN
15	10429.5	99.4	2016	4	AA882241 Human SCN
16	10428.5	99.4	2016	4	AA882244 Human SCN
17	10427.5	99.4	2016	4	AA882240 Human SCN
18	10425.5	99.4	2016	4	AA882243 Human SCN
19	10421.5	99.4	2016	2	AAW23994 Human htl
20	10416	99.3	2015	4	AA882242 Human SCN
21	9767	93.1	2019	4	AA67913 Cardiac s
22	9719	92.7	2020	2	AA806584 Cardiac s
23	7659.5	73.0	1603	4	AAU19518 Human dia

24	6406.5	61.1	2000	8	ADP79541 Human sod
25	6394.5	61.0	2005	4	AA899676 Human adu
26	6394.5	61.0	2005	9	ADY27148 Human SCN
27	6393.5	61.0	2005	9	ADY27149 Human SCN
28	6392.5	60.9	2005	5	AB83627 Human GE
29	6392.5	60.9	2005	7	AB878604 Human sod
30	6391.5	60.9	2005	7	AD878603 Human sod
31	6391.5	60.9	2005	7	AD878605 Human sod
32	6391.5	60.9	2005	7	AD878605 Human sod
33	6391.5	60.9	2005	8	AD878605 Human sod
34	6391.5	60.9	2005	8	AD878605 Human sod
35	6390.5	60.9	2005	4	ADY27150 Human SCN
36	6389.5	60.9	2005	4	AA899677 Human neo
37	6387.5	60.8	2005	9	ADY27147 Human SCN
38	6377.5	60.8	2000	5	AB806027 Human sod
39	6377.5	60.8	2000	8	AD878622 Human adu
40	6377.5	60.8	2000	8	ADP79545 Human sod
41	6377.5	60.8	2000	9	AA844248 Human sod
42	6342.5	60.5	1979	9	AA844245 Human sod
43	6329.5	60.3	2009	4	AA899674 Human adu
44	6329.5	60.3	2009	7	AD883180 Human SCN
45	6329.5	60.3	2009	7	AD883180 Human SCN
46	6327.5	60.3	2009	7	AD883180 Human SCN
47	6327.5	60.3	2009	7	AD883180 Human SCN
48	6324.5	60.3	2009	5	AA820515 Human ion
49	6324.5	60.3	2009	5	AB883626 Human GE
50	6320.5	60.3	2009	8	AD887512 Human sod
51	6319.5	60.2	2009	5	AB869292 Human sod
52	6319	60.2	1998	7	AB883184 Human SCN
53	6319	60.2	1998	7	AD826269 Human SCN
54	6319	60.2	1998	9	AE843184 Human pot
55	6317.5	60.2	2009	8	AD887505 Human sod
56	6316.5	60.2	2009	5	AB869291 Human sod
57	6316.5	60.2	2009	5	AB869290 Human tra
58	6316.5	60.2	2009	5	AA816776 Human tra
59	6316.5	60.2	2009	7	AD878598 Human sod
60	6316.5	60.2	2009	8	AD878509 Human sod
61	6315.5	60.2	2009	8	AD887515 Human sod
62	6315.5	60.2	2009	8	AD887513 Human sod
63	6315.5	60.2	2009	8	AD887532 Human sod
64	6315.5	60.2	2009	8	AD887507 Human sod
65	6314.5	60.2	2009	7	AD878593 Human sod
66	6314.5	60.2	2009	8	AD887508 Human sod
67	6314.5	60.2	2009	8	AD887511 Human sod
68	6314.5	60.2	2009	8	AD887504 Human sod
69	6314.5	60.2	2009	8	AD887530 Human sod
70	6314.5	60.2	2009	9	ADY27139 Human SCN
71	6314	60.2	1998	5	AA820510 Human ion
72	6313.5	60.2	2009	7	AD878595 Human sod
73	6313.5	60.2	2009	8	AD887506 Human sod
74	6313.5	60.2	2009	9	ADY27142 Human SCN
75	6313.5	60.2	2009	9	ADY27140 Human SCN
76	6313.5	60.2	2009	9	ADY27144 Human SCN
77	6312.5	60.2	2009	5	AB869293 Human sod
78	6312.5	60.2	2009	7	AD878599 Human sod
79	6312.5	60.2	2009	8	AD887514 Human sod
80	6311.5	60.2	2009	8	AD887503 Human sod
81	6310.5	60.2	2009	5	AB869289 Human sod
82	6310.5	60.2	2009	8	AD887502 Human sod
83	6310.5	60.2	2009	8	AD887531 Human sod
84	6310.5	60.2	2009	9	ADY27141 Human SCN
85	6309.5	60.2	2009	7	AD878594 Human sod
86	6308.5	60.1	1990	7	AD878607 Human sod
87	6308	60.1	1951	8	ADP79543 Human sod
88	6307.5	60.1	2009	8	AD887510 Human sod
89	6300.5	60.1	1999	5	AB806026 Human sod
90	6300.5	60.1	1999	4	ADU78366 Human vol
91	6289	60.0	1951	4	AA899678 Human adu
92	6287	59.9	1973	5	AA820516 Human ion
93	6285.5	59.9	1981	7	AB883185 Human SCN
94	6283	59.9	1951	4	AA899679 Human neo
95	6281	59.9	1951	8	ADL06576 Human tum
96	6281	59.9	1951	9	AA844250 Human SCN

97	6276.5	59.8	1962	5	AAE20511	AAE20511 Human ion
98	6272.5	59.8	1951	7	AAE59628	AAE59628 Rat Prote
99	6272.5	59.8	1951	7	AAE44252	AAE44252 Rat sodiu
100	6257.5	59.7	1956	4	AAE65785	AAE65785 Human SNS
101	6257.5	59.7	1956	4	AAE61996	AAE61996 Human per
102	6257.5	59.7	1956	6	ABG76193	ABG76193 Human vol
103	6257.5	59.7	1956	6	ABP72253	ABP72253 Human PN3
104	6257.5	59.7	1956	6	ADA50152	ADA50152 Human per
105	6257.5	59.7	1956	7	ADJ68903	ADJ68903 Human hea
106	6257.5	59.7	1956	8	ADJ68471	ADJ68471 Human Nav
107	6257.5	59.7	1956	8	ADU21150	ADU21150 Human sod
108	6257.5	59.7	1956	9	ADJ26252	ADJ26252 Novel cel
109	6257.5	59.7	1956	9	ABE90786	ABE90786 Human per
110	6253.5	59.6	1956	8	ADU21152	ADU21152 Human sod
111	6253.5	59.6	1956	6	ABG75945	ABG75945 Human per
112	6245	59.5	1984	7	ABE54547	ABE54547 Rat Prote
113	6245	59.5	1984	7	ABE54551	ABE54551 Rat Prote
114	6245	59.2	1984	7	ADJ63027	ADJ63027 Rat Prote
115	6213.5	59.2	1977	8	ADJ77202	ADJ77202 Human pro
116	6213.5	59.2	1977	8	ADL13028	ADL13028 Human ste
117	6213.5	59.2	1977	9	ABE86379	ABE86379 Amino aci
118	6213.5	59.2	1978	7	ABE54553	ABE54553 Human Pro
119	6213.5	59.2	1978	7	ABE54549	ABE54549 Human Pro
120	6212.5	59.2	1977	9	ABE86346	ABE86346 Amino aci
121	6212.5	59.2	1977	9	ABE86347	ABE86347 Amino aci
122	6212.5	59.2	1977	9	ABE86343	ABE86343 Amino aci
123	6212.5	59.2	1977	9	ABE86348	ABE86348 Amino aci
124	6210.5	59.2	1977	9	ABE86345	ABE86345 Amino aci
125	6207.5	59.2	1977	2	AAE9641	AAE9641 Periphra
126	6206	59.2	1978	2	AAE69361	AAE69361 Tetrodoto
127	6205.5	59.2	1977	9	ABE86344	ABE86344 Amino aci
128	6198	59.1	1978	9	ADJ26338	ADJ26338 Novel cel
129	6197	59.1	1980	9	ADJ28372	ADJ28372 Human SCN
130	6197	59.1	1980	2	AAE69362	AAE69362 Tetrodoto
131	6194	59.1	1980	7	ADJ78600	ADJ78600 Human sod
132	6194	59.1	1984	7	AAE96639	AAE96639 Periphra
133	6189	59.0	1980	7	ADJ78606	ADJ78606 Human sod
134	6186	59.0	1989	2	AAE92317	AAE92317 Periphra
135	6178	58.9	1942	8	ADJ87534	ADJ87534 Mutant SC
136	6140	58.5	1980	3	AAE23563	AAE23563 Human sod
137	6140	58.5	1980	5	AAO14927	AAO14927 Human sod
138	6138.5	58.5	1962	2	AAV17250	AAV17250 NaNg poly
139	6110.5	58.3	1942	8	ADU21164	ADU21164 Guinea pi
140	6103.5	58.2	1957	6	ADA50156	ADA50156 Rat perip
141	6100	58.2	1956	4	AAE65783	AAE65783 Rat SNS1
142	6100	58.2	1956	4	AAE61995	AAE61995 Rat perip
143	6100	58.2	1956	6	ADA50153	ADA50153 Rat perip
144	6100	58.2	1956	8	ADU21165	ADU21165 Rat sodiu
145	6100	58.2	1956	9	ABE90788	ABE90788 Rat perip
146	6088.5	58.0	1957	2	AAW21740	AAW21740 Variant r
147	6087.5	58.0	1957	6	ABG76191	ABG76191 Rat volta
148	6087.5	58.0	1957	7	ADD44754	ADD44754 Rat Prote
149	6087.5	58.0	1957	8	ADJ08469	ADJ08469 Rat Nav 1
150	6087.5	58.0	1957	8	ADU21148	ADU21148 Rat sodiu
151	6087.5	58.0	1957	8	ADJ26396	ADJ26396 Novel cel
152	6086.5	58.0	1957	2	AAW21737	AAW21737 Wild type
153	6078	57.9	1891	9	ADJ27145	ADJ27145 Human SCN
154	6078	57.9	1958	4	AAE65784	AAE65784 Mouse SNS
155	6066.5	57.8	1836	7	ADJ26323	ADJ26323 Novel cel
156	6066.5	57.8	1836	7	ADJ57388	ADJ57388 Human Pro
157	6066.5	57.8	1836	7	ADJ59630	ADJ59630 Human Pro
158	6066.5	57.8	1836	7	ADJ63029	ADJ63029 Human Pro
159	6060.5	57.8	1836	8	ADJ17412	ADJ17412 Human sof
160	6059.5	57.8	1836	8	AEF53825	AEF53825 Human vol
161	6049.5	57.7	1957	6	ADA50155	ADA50155 Rat perip
162	6049	57.7	1957	6	ADJ57386	ADJ57386 Rat Prote
163	6046	57.6	1956	6	ABG75944	ABG75944 Rat perip
164	6046	57.6	1956	6	ADA50144	ADA50144 Rat perip
165	6046	57.6	1956	6	ABE90778	ABE90778 Rat perip
166	6043	57.6	2132	2	AAW21739	AAW21739 Variant r
167	6012	57.3	1989	2	AAE96640	AAE96640 Periphra
168	5876	56.0	1855	7	ADJ78597	ADJ78597 Human sod
169	5644	53.8	1795	7	ADJ78596	ADJ78596 Human sod

170	5566.5	53.1	1835	2	AAE92316	AAE92316 Periphra
171	5456	52.0	1107	6	ABR41495	ABR41495 Human DIT
172	5293	50.5	1726	2	ADJ288370	ADJ288370 Human SCN
173	4811	45.9	1765	2	AAE05597	AAE05597 Mouse sod
174	4811	45.9	1765	4	AAE20124	AAE20124 Mouse sod
175	4811	45.9	1765	4	AAE20126	AAE20126 Mouse Na
176	4811	45.9	1765	7	ADJ26317	ADJ26317 Novel cel
177	4811	45.9	1765	9	AAO14925	AAO14925 Human sod
178	4729	45.1	1791	5	AAO14925	AAO14925 Novel cel
179	4729	45.1	1825	9	ADJ26245	ADJ26245 Human SCN
180	4728	45.1	1765	7	ADJ26219	ADJ26219 Rat Na v
181	4728	45.1	1765	7	ADJ26210	ADJ26210 Rat Na v
182	4728	45.1	1765	7	ADJ26210	ADJ26210 Rat Prote
183	4728	45.1	1765	9	ADJ26389	ADJ26389 Novel cel
184	4724	45.0	1765	2	AAE16572	AAE16572 Type 5 so
185	4724	45.0	1791	4	AAE20121	AAE20121 Human sod
186	4724	45.0	1791	7	ADJ26319	ADJ26319 Human Na
187	4720	45.0	1765	2	AAE06596	AAE06596 Rat sodiu
188	4720	45.0	1765	4	AAE20122	AAE20122 Rat sodiu
189	4720	45.0	1765	4	AAE20123	AAE20123 Rat sodiu
190	4719	45.0	1765	2	AAE16668	AAE16668 Rat senso
191	4711	44.9	1753	9	ADJ26297	ADJ26297 Human SCN
192	4488.5	42.8	1510	8	ADJ87522	ADJ87522 Mutant SC
193	4207.5	40.1	1453	5	AAE20512	AAE20512 Human ion
194	4197	40.0	1442	5	AAE20512	AAE20512 Human ion
195	4173.5	39.8	2131	8	ADJ64743	ADJ64743 Drosophi1
196	4173.5	39.8	2131	8	ADJ30141	ADJ30141 Drosophi1
197	4169.5	39.8	2131	8	ADJ30146	ADJ30146 Drosophi1
198	4167	39.7	2105	2	AAE57772	AAE57772 Musca dom
199	4167	39.7	2105	2	AAE89577	AAE89577 Calci1um p
200	4162	39.7	2104	2	AAE57773	AAE57773 Musca dom

ALIGNMENTS

RESULT 1

ADJ34001

ID ADJ34001 standard; protein: 2015 AA.

XX AC ADJ34001;

XX DT 03-JUN-2004 (first entry)

XX DE Human SCN α variant 4 protein SEQ ID NO:8.

XX KW human; cardiac sodium channel alpha subunit; SCN5A;

XX KW sodium voltage-gated channel; type V; alpha; mutation study;

XX KW sodium channel related disease.

XX OS Homo sapiens.

XX WO2004012668-A2.

XX 12-FEB-2004.

XX PD 01-AUG-2003; 2003WO-US024190.

XX PE 02-AUG-2002; 2002US-0401018P.

XX PR (WISC) WISCONSIN ALUMNI RES FOUND.

XX PA Makieleki JC, Ye B, Ackerman MJ;

XX PI WPI; 2004-157000/15.

XX DR N-PSDB; ADJ34000.

XX PT New nucleic acid encoding sodium voltage-gated channel, type V, alpha

XX PT polypeptide variants, useful for studying mutations, and designing or

XX PT identifying new diagnostics and treatment strategies or agents for sodium

XX PT channel related diseases.

XX PS Claim 17; SEQ ID NO 8; 111pp; English.

Same

XX The present sequence represents a variant of the human cardiac sodium channel alpha subunit designated SCN5A (sodium voltage-gated channel, type V, alpha) protein. The present invention describes an isolated polynucleotide encoding an SCN5A polypeptide where the polynucleotide is selected from the group: (1) a first polynucleotide that encodes an SCN5A polypeptide selected from the group consisting of: (i) a histidine, threonine, leucine, arginine and glutamine at amino acid positions 558, 559, 618, 1027 and 1077, respectively; (ii) an arginine, threonine, leucine, arginine and glutamine at amino acid positions 558, 559, 618, 1027 and 1077, respectively; (iii) an arginine, threonine, leucine, arginine and glutamine at amino acid positions 558, 559, 618, 1027 and 1077, respectively; (iv) an arginine, threonine, leucine, arginine and glutamine at amino acid positions 558, 559, 618 and 1027, respectively, with the amino acid at amino acid position 1077 deleted; (2) a second polynucleotide that is at least 80 % identical to the first polynucleotide over the entire length of the first polypeptide; (3) a third polynucleotide that encodes any of the SCN5A polypeptides with a conservative substitution, deletion or rearrangement at one or more non-critical amino acid position; and (4) a fourth polynucleotide that is a complement of the first, second or third polynucleotide. Also described: (1) a genetic construct comprising the polynucleotide operably linked to a non-native expression control sequence; (2) a cell comprising the polynucleotide; (3) an isolated polypeptide encoded by the polynucleotide; (4) an antibody that specifically binds to the polypeptide; (5) identifying an agent that can alter the activity of a sodium channel; (6) identifying an agent that can alter the expression of a sodium channel; (7) determining whether a biological sample or a preparation derived from the biological sample contains the polypeptide; (8) determining whether a mutation on a sodium channel is associated with a disease; and (9) determining whether a human or non-human subject is at risk for long QT syndrome. The SCN5A polynucleotides and polypeptides are useful for studying mutations, and designing or identifying new diagnostics and treatment strategies or agents for sodium channel related diseases or conditions.

Sequence 2015 AA;

Query Match 100.0%; Score 10489; DB 8; Length 2015;
Beet Local Similarity 100.0%; Pred. No. 0;
Matches 2015; Conservative 0; Mismatches 0; Indels 0; Gaps 0;

QY 1 MANFLPRGTSFRRFRESLAIEKMAEKQARGSTTLOESREGPEEAREPQLODA 60
DB 1 MANFLPRGTSFRRFRESLAIEKMAEKQARGSTTLOESREGPEEAREPQLODA 60
QY 61 SKKLDLYGNPQELIGBELEDLPFYSTQKTFIVLNGKTIIFRSATNALVLSPPHPI 120
DB 61 SKKLDLYGNPQELIGBELEDLPFYSTQKTFIVLNGKTIIFRSATNALVLSPPHPI 120
QY 121 RRAAKVILVHSLFNNLIMCTIITNCVMAQHDPPMTKYVEYTFPAIYFESLVYILARG 180
DB 121 RRAAKVILVHSLFNNLIMCTIITNCVMAQHDPPMTKYVEYTFPAIYFESLVYILARG 180
QY 181 FCLHAFTFLRDPMMNLDFSIIIMATTEFVLDGANSALRTFPRVLAALKTISVIGKTIIV 240
DB 181 FCLHAFTFLRDPMMNLDFSIIIMATTEFVLDGANSALRTFPRVLAALKTISVIGKTIIV 240
QY 241 GALIGSVKKLADVWLVTFCLSVFALIGLQFMGNLRHKCVANFTALNGTNGSVADGLV 300
DB 241 GALIGSVKKLADVWLVTFCLSVFALIGLQFMGNLRHKCVANFTALNGTNGSVADGLV 300
QY 301 WESLDLYSDPENNYLLKNGTSVLLCGNSSDAGTCPEGRCLKAENPDHGTSTDSFAM 360
DB 301 WESLDLYSDPENNYLLKNGTSVLLCGNSSDAGTCPEGRCLKAENPDHGTSTDSFAM 360
QY 361 AFLALFRMTODCWERLYQOOLRSAGKTYMIFPMLVIFLGSFYLVNLLAVAAAYEEN 420
DB 361 AFLALFRMTODCWERLYQOOLRSAGKTYMIFPMLVIFLGSFYLVNLLAVAAAYEEN 420
QY 421 QATIAETEKEKRRFOEAMEMLKKEHEALTIRGVDTVSSRSLSMSPLAPNSHERSKRK 480
DB 421 QATIAETEKEKRRFOEAMEMLKKEHEALTIRGVDTVSSRSLSMSPLAPNSHERSKRK 480

QY 481 RMSSGTECEGDRLPKSDSEDPGRAMNHLSTRGLSRTSMKRSSRGSIFFTRRRDLGSE 540
DB 481 RMSSGTECEGDRLPKSDSEDPGRAMNHLSTRGLSRTSMKRSSRGSIFFTRRRDLGSE 540
QY 541 ADPADENSTAGESSHRTSLVWPLRRTSAQGPSPTGSAAGHALHGNKSTVDCNGV 600
DB 541 ADPADENSTAGESSHRTSLVWPLRRTSAQGPSPTGSAAGHALHGNKSTVDCNGV 600
QY 601 VSLGADPEATSPGSHLRPMLHPPPTTTPSEPGGPQULTSQAPCVDFEFGAQQ 660
DB 601 VSLGADPEATSPGSHLRPMLHPPPTTTPSEPGGPQULTSQAPCVDFEFGAQQ 660
QY 661 RALSASVLTSLALEBSRHKCPCCMRRLAORYLIWECCPLMMSIKQGVKLVMDPFD 720
DB 661 RALSASVLTSLALEBSRHKCPCCMRRLAORYLIWECCPLMMSIKQGVKLVMDPFD 720
QY 721 LITIMCIYVNLTFMALHYNMTSFEEMLOVGNLFTFTGIFTAEMTFKIIALDPYFYFOG 780
DB 721 LITIMCIYVNLTFMALHYNMTSFEEMLOVGNLFTFTGIFTAEMTFKIIALDPYFYFOG 780
QY 781 WNIPIISIIIVLSLMEIGLSRMSNLSVLRSPRLRYFKLAKSWPTNLTKIIGNSVGALG 840
DB 781 WNIPIISIIIVLSLMEIGLSRMSNLSVLRSPRLRYFKLAKSWPTNLTKIIGNSVGALG 840
QY 841 NLTIVLATTIIFPAVVGMOIRGKNYSELSDSGILPPMHMDPFAHPIIRIICGEMI 900
DB 841 NLTIVLATTIIFPAVVGMOIRGKNYSELSDSGILPPMHMDPFAHPIIRIICGEMI 900
QY 901 ETMDMCEVSGSLCLFVLLVMVIGNLVNLFPLALISFSASDNLTPADERREMNLIQ 960
DB 901 ETMDMCEVSGSLCLFVLLVMVIGNLVNLFPLALISFSASDNLTPADERREMNLIQ 960
QY 961 LALARIQGLRFVKTITWDPCGGLRQRPQKPAALAAQOLPSCITATYSPPEPETEKP 1020
DB 961 LALARIQGLRFVKTITWDPCGGLRQRPQKPAALAAQOLPSCITATYSPPEPETEKP 1020
QY 1021 PTRKREPEBEOPOGPRPDPEPCVPIAAVESTDDOEBEENSILGTEBESSKQESOP 1080
DB 1021 PTRKREPEBEOPOGPRPDPEPCVPIAAVESTDDOEBEENSILGTEBESSKQESOP 1080
QY 1081 VSGGEAPDPDSRTMSQVSATASSEBAASASQADMRQMAEPOAGCETPEBSCSEGT 1140
DB 1081 VSGGEAPDPDSRTMSQVSATASSEBAASASQADMRQMAEPOAGCETPEBSCSEGT 1140
QY 1141 ADMNTAELBQIPDLGQDVXDPEDCFTGECVRRCPCCAVDTTQAPGKVMRLRTCYHI 1200
DB 1141 ADMNTAELBQIPDLGQDVXDPEDCFTGECVRRCPCCAVDTTQAPGKVMRLRTCYHI 1200
QY 1201 VEHWSFEFTIIFMILLSSGALAFEDIYLBERTIKVLEEVADKMTYVFVEMLLKWAY 1260
DB 1201 VEHWSFEFTIIFMILLSSGALAFEDIYLBERTIKVLEEVADKMTYVFVEMLLKWAY 1260
QY 1261 GFKKYFTNAKCMLEPLVDVSLVANTLFAEWGPIKSLRTLRALPLRLASFEGMR 1320
DB 1261 GFKKYFTNAKCMLEPLVDVSLVANTLFAEWGPIKSLRTLRALPLRLASFEGMR 1320
QY 1321 VVNAALVGAISIMNVLLVCLIFMLIFSIMGVNLPAKRGRCINOTEGDPLNNTIYNNK 1380
DB 1321 VVNAALVGAISIMNVLLVCLIFMLIFSIMGVNLPAKRGRCINOTEGDPLNNTIYNNK 1380
QY 1381 SOCESLNTLGEFLYTKVNFNDNGAGYALALQVATPFQNMWDIWAADSDSGYEQPOME 1440
DB 1381 SOCESLNTLGEFLYTKVNFNDNGAGYALALQVATPFQNMWDIWAADSDSGYEQPOME 1440
QY 1441 YNLWYIYFVFIIFGSEFTNLPLGVIIDNFNOOKKGLGGODIFMTEBQKXYNAMKGL 1500
DB 1441 YNLWYIYFVFIIFGSEFTNLPLGVIIDNFNOOKKGLGGODIFMTEBQKXYNAMKGL 1500
QY 1501 GSKKPQKPIPPPLNKYQGFIDIVTKQAFDVTIMFLICLNNTVMVETDDOSPDKINILA 1560
DB 1501 GSKKPQKPIPPPLNKYQGFIDIVTKQAFDVTIMFLICLNNTVMVETDDOSPDKINILA 1560

QY 1561 KINLLPVAIFPGEICVLAARHYFTNSMNIPOVWVUUSVIGTVLSDIIQKXFESEPTL 1620
DB 1561 KINLLPVAIFPGEICVLAARHYFTNSMNIPOVWVUUSVIGTVLSDIIQKXFESEPTL 1620
QY 1621 FRVRLARIGILRLIRGAKGIRTLTFLMMSLPALFNIGLLFLVMEYISIFGMANPAY 1680
DB 1621 FRVRLARIGILRLIRGAKGIRTLTFLMMSLPALFNIGLLFLVMEYISIFGMANPAY 1680
QY 1681 VKWAGIDDMFNPOTFANSMLCFQITTSAGMDGILSPINTGPPYCDPLPNSNGSRGD 1740
DB 1681 VKWAGIDDMFNPOTFANSMLCFQITTSAGMDGILSPINTGPPYCDPLPNSNGSRGD 1740
QY 1741 CGSPAVGLFFFTYIIISFLVWVNYIAIILENSVAEESTEPSEDDPMFEIMEKF 1800
DB 1741 CGSPAVGLFFFTYIIISFLVWVNYIAIILENSVAEESTEPSEDDPMFEIMEKF 1800
QY 1801 DPEATQFIEYSVLSPFADALSEPLRIAKPNOISLINMDLPVWSGDRICMIDILFAFTKRV 1860
DB 1801 DPEATQFIEYSVLSPFADALSEPLRIAKPNOISLINMDLPVWSGDRICMIDILFAFTKRV 1860
QY 1861 LGESGEMDALKIOMEKEMANPSKISYEPIITTLRRHGEVSAMVIOQAFRRHLLQPSL 1920
DB 1861 LGESGEMDALKIOMEKEMANPSKISYEPIITTLRRHGEVSAMVIOQAFRRHLLQPSL 1920
QY 1921 KHAStLFRQAGSGISEEDABEREGLIAVWMSNSRPLGPPSSSISSTSPPSYDSVT 1980
DB 1921 KHAStLFRQAGSGISEEDABEREGLIAVWMSNSRPLGPPSSSISSTSPPSYDSVT 1980
QY 1981 RATSNDLQVRGSDYSHSEDLADFPSPDRRESIV 2015
DB 1981 RATSNDLQVRGSDYSHSEDLADFPSPDRRESIV 2015

RESULT 2
ADFS6441
ID ADFS6441 standard; protein; 2015 AA.

AC ADFS6441;

DT 12-FEB-2004 (first entry)

DE Human Nav1.5 sodium channel alpha subunit SCN5A h1b.

KM cardiac; antiarrhythmic; sodium channel blocker;

KW sodium channel activator; SCN5A channel blocker; SCN5A channel activator;

KW sodium channel subunit h1b; h1b; SCN5A; long QT syndrome;

KW sodium channel; cardiac arrhythmia; heart disorder;

KW human Nav1.5 sodium channel alpha subunit SCN5A.

OS Homo sapiens.

PN US2003157600-A1.

PD 21-AUG-2003.

PF 12-FEB-2002; 2002US-00077054.

PR 12-FEB-2002; 2002US-00077054.

PA (MAKI/) MAKIELSKI J C.

PI (YEBB/) YE B.

PI Makieleki JC, Ye B;

DR MPI; 2003-688984/65.

DR N-PESDB; ADFS6440.

XX New sodium channel subunit h1b polypeptide, useful for screening for

PT compounds which alter sodium channel activity for treating cardiac

PT arrhythmia and other sodium channel related cardiac conditions.

XX Claim 1; SEQ ID NO 2; 24pp; English.

CC The invention describes an isolated sodium channel subunit h1b
CC polypeptide (I) comprising a fully defined 2015 amino acid sequence as
CC given in the specification optionally, carrying a conservative
CC substitution, deletion or rearrangement at one or more non-critical amino
CC acid position. Detecting the presence of the h1b SCN5A gene is useful
CC for determining if a (non-)human subject is at risk for Long QT syndrome.
CC Agents screened that increase or decrease sodium channel activity are
CC useful for treating cardiac arrhythmia and other sodium channel related
CC heart disorders. This is the amino acid sequence of human Nav1.5 sodium
CC channel alpha subunit SCN5A.

XX Sequence 2015 AA;

Query Match 4 99.9%; Score 10487; DB 7; Length 2015;
Best Local Similarity 99.9%; Pred. No. 0;
Matches 2014; Conservative 1; Mismatches 0; Indels 0; Gaps 0;

QY 1 MANFLPRTGSPRRFRRESIAAIEKMAEKQARGSTTLQESREGLEPEEAPRQDLQA 60
DB 1 MANFLPRTGSPRRFRRESIAAIEKMAEKQARGSTTLQESREGLEPEEAPRQDLQA 60
QY 61 SKKLPLDYGNDPPELICEPLEDDLPFYSTKTFIVLKKGTIRFSATNALYVLSPPHP 120
DB 61 SKKLPLDYGNDPPELICEPLEDDLPFYSTKTFIVLKKGTIRFSATNALYVLSPPHP 120
QY 121 RRAAVKILVHSLFNMILMCTILNVCVMAQDPPPTKYVEYFTAIYFESLVKILARG 180
DB 121 RRAAVKILVHSLFNMILMCTILNVCVMAQDPPPTKYVEYFTAIYFESLVKILARG 180
QY 181 FCLHAFPLDPMWMDLDFSVIIMAYTTEFDLGVSAALRFRVLRAKLTISVIGLKTIV 240
DB 181 FCLHAFPLDPMWMDLDFSVIIMAYTTEFDLGVSAALRFRVLRAKLTISVIGLKTIV 240
QY 241 GALIQSVKCLADVWVTFVCLSVFALIGLQFPMKRLHKCVRNPTALNGNSVEADGLV 300
DB 241 GALIQSVKCLADVWVTFVCLSVFALIGLQFPMKRLHKCVRNPTALNGNSVEADGLV 300
QY 241 GALIQSVKCLADVWVTFVCLSVFALIGLQFPMKRLHKCVRNPTALNGNSVEADGLV 300
DB 241 GALIQSVKCLADVWVTFVCLSVFALIGLQFPMKRLHKCVRNPTALNGNSVEADGLV 300
QY 301 WESLDLYLSPDENYLNKGTSDVLLCGNSSDAGCPGEGYCLKAGENDHGYTSFDSFAW 360
DB 301 WESLDLYLSPDENYLNKGTSDVLLCGNSSDAGCPGEGYCLKAGENDHGYTSFDSFAW 360
QY 361 AFLALFRLMTQDCWERYLQOTLRGAKIYMFMLVIFLGSFYLVNLLAVVMAAYEON 420
DB 361 AFLALFRLMTQDCWERYLQOTLRGAKIYMFMLVIFLGSFYLVNLLAVVMAAYEON 420
QY 421 QATTAETBEKRRQEMEMLKKEHEALTTRGVDTVSRSLSLEMSPLAPVNSHERSRK 480
DB 421 QATTAETBEKRRQEMEMLKKEHEALTTRGVDTVSRSLSLEMSPLAPVNSHERSRK 480
QY 481 RMSGTECEGDRLPKSDSEDPGRAMNHLSTRGLSRSMKPRSSRSISFTFRRDIGSE 540
DB 481 RMSGTECEGDRLPKSDSEDPGRAMNHLSTRGLSRSMKPRSSRSISFTFRRDIGSE 540
QY 541 ADPADENSTAGSESHRTSLVWPPLRTSAQOQSPGTSAPGHALHGKKNSTVDCNV 600
DB 541 ADPADENSTAGSESHRTSLVWPPLRTSAQOQSPGTSAPGHALHGKKNSTVDCNV 600
QY 601 VSLIAGDPEATSPGSHILRPWMLRHPDPTTPEEBEGGQMLTSQAPCVUDGFEERARQ 660
DB 601 VSLIAGDPEATSPGSHILRPWMLRHPDPTTPEEBEGGQMLTSQAPCVUDGFEERARQ 660
QY 661 RALSAVSVTSLALELESRRHKCPCKNRLAQRILIMECCPLMMSIQGVLYVMDPFTD 720
DB 661 RALSAVSVTSLALELESRRHKCPCKNRLAQRILIMECCPLMMSIQGVLYVMDPFTD 720
QY 721 LITMCIIVLNTLPMALRHYNTSEFEBMLQVGNLVFTGIFTAEMTFKIALDPYYRQOG 780
DB 721 LITMCIIVLNTLPMALRHYNTSEFEBMLQVGNLVFTGIFTAEMTFKIALDPYYRQOG 780
QY 781 WNIIDSIIVILSLMELGSRMSNLSVLRSPFLRVLAVFKLAKSWPTLNTLIKIGSVGALG 840
DB 781 WNIIDSIIVILSLMELGSRMSNLSVLRSPFLRVLAVFKLAKSWPTLNTLIKIGSVGALG 840

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Qy 841 NLTVLVAIVFI FAVVGMQLFGKNYSELSDSGLLPRWMMDFFAHFIIRILGEMI 900
    |||
Db 841 NLTVLVAIVFI FAVVGMQLFGKNYSELSDSGLLPRWMMDFFAHFIIRILGEMI 900
Qy 901 ETTMDCMEVSGSLCLVFLVWVIGNLVNLFLALLSSFSADNLTADEDEMNLIQ 960
    |||
Db 901 ETTMDCMEVSGSLCLVFLVWVIGNLVNLFLALLSSFSADNLTADEDEMNLIQ 960
Qy 961 LALARIQRLRFRVYKRTTNPFCGGLRQRQKPAALAAOQLPSCIA TPSPPEPEKXP 1020
    |||
Db 961 LALARIQRLRFRVYKRTTNPFCGGLRQRQKPAALAAOQLPSCIA TPSPPEPEKXP 1020
Qy 1021 PTKETRFEEGEPQGGTGPDEPVCVPIAVASDTPDEBEBENSIGTEBESSKQESOP 1080
    |||
Db 1021 PTKETRFEEGEPQGGTGPDEPVCVPIAVASDTPDEBEBENSIGTEBESSKQESOP 1080
Qy 1081 VSGGPAPPSRTWSQVATASSEASASQADMROQWAFPOAPCGETPEDSCSGEST 1140
    |||
Db 1081 VSGGPAPPSRTWSQVATASSEASASQADMROQWAFPOAPCGETPEDSCSGEST 1140
Qy 1141 ADMTTAELEBQIPDLGQDVDPEDCGTCYRCCCAVDTTQAPGKYMRLKTCYHI 1200
    |||
Db 1141 ADMTTAELEBQIPDLGQDVDPEDCGTCYRCCCAVDTTQAPGKYMRLKTCYHI 1200
Qy 1201 VEHSMFEFPIIFMILLSSGALAPBDIYLBERTIKVLEADKMFTYVFLMILKMYAY 1260
    |||
Db 1201 VEHSMFEFPIIFMILLSSGALAPBDIYLBERTIKVLEADKMFTYVFLMILKMYAY 1260
Qy 1261 GFKKFTTNAMCWLDFLVDVSLVSVANTLGAEMGPISKSLRTLRLALRALSPFEGNR 1320
    |||
Db 1261 GFKKFTTNAMCWLDFLVDVSLVSVANTLGAEMGPISKSLRTLRLALRALSPFEGNR 1320
Qy 1321 VVWNLVGAIPBSIMNVLVCLIFMLIFSMGVNLFAGKRGRCINOTEGDPLANTYIVNKK 1380
    |||
Db 1321 VVWNLVGAIPBSIMNVLVCLIFMLIFSMGVNLFAGKRGRCINOTEGDPLANTYIVNKK 1380
Qy 1381 SOCESLNTGELYMTKVKVNFNVGAGYALLQVATPFKMMIMYAAVDSRGYEBOPOME 1440
    |||
Db 1381 SOCESLNTGELYMTKVKVNFNVGAGYALLQVATPFKMMIMYAAVDSRGYEBOPOME 1440
Qy 1441 YNLWYIYFVFIIFGSPFTLNLFTGVIIIDNFOOKKLGODIMTEBOKKYANAMKUL 1500
    |||
Db 1441 YNLWYIYFVFIIFGSPFTLNLFTGVIIIDNFOOKKLGODIMTEBOKKYANAMKUL 1500
Qy 1501 GSKKQKQKPRPIANKYQGFIPDIVTKQAFDVTIMELICLNMVTMWVEDTDQSPKINILIA 1560
    |||
Db 1501 GSKKQKQKPRPIANKYQGFIPDIVTKQAFDVTIMELICLNMVTMWVEDTDQSPKINILIA 1560
Qy 1561 KINLFFVAIFGECIVKLAALAHYYFTNSWNI FDFVVVLTISVGTVLSDIIOKXFPSPTL 1620
    |||
Db 1561 KINLFFVAIFGECIVKLAALAHYYFTNSWNI FDFVVVLTISVGTVLSDIIOKXFPSPTL 1620
Qy 1621 FRVIRLARIGRILIRGAKGIRTLIFALMMSLPALFNIGLLFLVMFYISFGANAPAY 1680
    |||
Db 1621 FRVIRLARIGRILIRGAKGIRTLIFALMMSLPALFNIGLLFLVMFYISFGANAPAY 1680
Qy 1661 VKMEAGIDMNFQTFPANSMLCLFOITTSAGMDGLSPILNTGPPYCDPTLPNSGSGRD 1740
    |||
Db 1661 VKMEAGIDMNFQTFPANSMLCLFOITTSAGMDGLSPILNTGPPYCDPTLPNSGSGRD 1740
Qy 1741 CGSPAVGLIFFTTYIISFLIVVMNYIAIILENFVATEBESPELSDPDMFYIWEKF 1800
    |||
Db 1741 CGSPAVGLIFFTTYIISFLIVVMNYIAIILENFVATEBESPELSDPDMFYIWEKF 1800
Qy 1801 DPEATQFLEYSVLSDPADLSEPLRIAKPNQISLINMOLPMWSGRIRHOMDILFAFTRKV 1860
    |||
Db 1801 DPEATQFLEYSVLSDPADLSEPLRIAKPNQISLINMOLPMWSGRIRHOMDILFAFTRKV 1860
Qy 1861 LGESSEMDALKIQMEKPMANPSKISYEPITTTLRKHGEVSANVIOAPFRHLLQSSL 1920
    |||
Db 1861 LGESSEMDALKIQMEKPMANPSKISYEPITTTLRKHGEVSANVIOAPFRHLLQSSL 1920
Qy 1921 KHASFLPQOAGSGISEDAPEREGLIAVWSENSESRPLGPPSSSSISTSPSPYDSVT 1980
    |||

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Db 1921 KHASFLPQOAGSGISEDAPEREGLIAVWSENSESRPLGPPSSSSISTSPSPYDSVT 1980
    |||
Qy 1981 RATSNDLVQVSGSDYSHSEDLADFPSPDPDRRESIV 2015
    |||
Db 1981 RATSNDLVQVSGSDYSHSEDLADFPSPDPDRRESIV 2015
    |||

RESULT 3
ADM3399
ID ADM3399 standard; protein; 2015 AA.
XX
AC ADM3399;
XX
DT 03-JUN-2004 (first entry)
XX
DE Human SCN5A variant 3 protein SEQ ID NO:6.
XX
KW human; cardiac sodium channel alpha subunit; SCN5A;
KW sodium channel-gated channel; type V; alpha; mutation study;
KW sodium channel related disease.
XX
OS Homo sapiens.
XX
PN MO2004012668-A2.
PD 12-FEB-2004.
XX
PF 01-AUG-2003; 2003WO-US024190.
XX
PR 02-AUG-2002; 2002US-0401018P.
XX
(WISC ) WISCONSIN ALUMNI RES FOUND.
PI Makieleki JC, Ye B, Ackerman MJ;
XX
DR MPI: 2004-157000/15.
XX
DR N-PESDB; ADM3398.
XX
PT New nucleic acid encoding sodium voltage-gated channel, type V, alpha
PT polypeptide variants, useful for studying mutations, and designing or
PT identifying new diagnostics and treatment strategies or agents for sodium
PT channel related diseases.
XX
PS Claim 14; SEQ ID NO 6; 111pp; English.
XX

The present sequence represents a variant of the human cardiac sodium
channel alpha subunit designated SCN5A (sodium voltage-gated channel,
type V, alpha) protein. The present invention describes an isolated
polynucleotide encoding an SCN5A polypeptide where the polynucleotide is
selected from the group: (1) a first polynucleotide that encodes an SCN5A
polypeptide selected from the group consisting of: (i) a histidine,
CC threonine, leucine, arginine and glutamine at amino acid positions 558,
CC 559, 618, 1027 and 1077, respectively; (ii) an arginine, threonine,
CC leucine, arginine and glutamine at amino acid positions 558, 559, 618,
CC 1027 and 1077, respectively; (iii) a histidine, threonine, leucine and
CC arginine at amino acid positions 558, 559, 618 and 1027, respectively,
CC with the amino acid at amino acid position 1077 deleted; or (iv) an
CC arginine, threonine, leucine and arginine at amino acid positions 558,
CC 559, 618 and 1027, respectively, with the amino acid at amino acid
CC position 1077 deleted; (2) a second polynucleotide that is at least 80 %
CC identical to the first polynucleotide over the entire length of the first
CC polypeptide; (3) a third polynucleotide that encodes any of the SCN5A
CC polypeptides with a conservative substitution, deletion or rearrangement
CC at one or more non-critical amino acid position; and (4) a fourth
CC polynucleotide that is a complement of the first, second or third
CC polynucleotide. Also described: (1) a genetic construct comprising the
CC polynucleotide operably linked to a non-native expression control
CC sequence; (2) a cell comprising the polynucleotide; (3) an isolated
CC polypeptide encoded by the polynucleotide; (4) an antibody that
CC specifically binds to the polypeptide; (5) identifying an agent that can
CC alter the activity of a sodium channel; (6) identifying an agent that can
CC alter the expression of a sodium channel; (7) determining whether a

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biological sample or a preparation derived from the biological sample contains the polypeptide; (8) determining whether a mutation on a sodium channel is associated with a disease; and (9) determining whether a human or non-human subject is at risk for long QT syndrome. The SCNSA polynucleotides and polypeptides are useful for studying mutations, and designing or identifying new diagnostics and treatment strategies or agents for sodium channel related diseases or conditions.

Sequence 2015 AA:

Query Match 99.9%; Score 10484; DB 8; Length 2015;
Best Local Similarity 99.9%; Pred. No. 0;
Matches 2014; Conservative 0; Mismatches 1; Indels 0; Gaps 0;

1 MANFLPRGTSFRFRFTRESLAIEKRMAEKQARGSTTLQESRGLPEEAPRQDLQA 60
1 MANFLPRGTSFRFRFTRESLAIEKRMAEKQARGSTTLQESRGLPEEAPRQDLQA 60
61 SKKLPDLGNPQELIGPELDLPDFYSTOKTFTLVNKGKTIFFRSATNALVLSPPPI 120
61 SKKLPDLGNPQELIGPELDLPDFYSTOKTFTLVNKGKTIFFRSATNALVLSPPPI 120
121 RRAAVKILVHSLFNNLIMCTILTNCFVMAQHPPEWTKYVEYTFATYTFESLVKILARG 180
121 RRAAVKILVHSLFNNLIMCTILTNCFVMAQHPPEWTKYVEYTFATYTFESLVKILARG 180
121 RRAAVKILVHSLFNNLIMCTILTNCFVMAQHPPEWTKYVEYTFATYTFESLVKILARG 180
181 FCLHAFTEFLRDPWNNLDFSVIIMAYTTEFVLDGNVSAIRTRVRALAKTISVIGLKTIV 240
181 FCLHAFTEFLRDPWNNLDFSVIIMAYTTEFVLDGNVSAIRTRVRALAKTISVIGLKTIV 240
241 GALTOSVKKLADVMVLTVFCISVFPALIGLOLFMGVLRHKCVRNFPALNGTNGSEADGLV 300
241 GALTOSVKKLADVMVLTVFCISVFPALIGLOLFMGVLRHKCVRNFPALNGTNGSEADGLV 300
301 WESLDLYSDPENNYLLKNGTSDVLLCGNSDAGTCEGYRCLKAGENDHGTSPDFAW 360
301 WESLDLYSDPENNYLLKNGTSDVLLCGNSDAGTCEGYRCLKAGENDHGTSPDFAW 360
361 AFALFRLMTODCWERLYOQTLRSAGKIYMFEMLVFLGSPYLVNLLAVANAYEON 420
361 AFALFRLMTODCWERLYOQTLRSAGKIYMFEMLVFLGSPYLVNLLAVANAYEON 420
421 QATTAEKEKERKPEAMEMLKKEHEALTIRGVTVSSLSMSPLAVNSHERSKRK 480
421 QATTAEKEKERKPEAMEMLKKEHEALTIRGVTVSSLSMSPLAVNSHERSKRK 480
481 RMSSTEECGEDRLPKSDESDGPRAMNHLSTRGLSRTSMKPRSSRGSIFTFRRRLDGE 540
481 RMSSTEECGEDRLPKSDESDGPRAMNHLSTRGLSRTSMKPRSSRGSIFTFRRRLDGE 540
541 ADPADENSTAGESSESHTSLVWPMLRRTSAQOGPSRGTSAFGALHGKKNSTYDCGV 600
541 ADPADENSTAGESSESHTSLVWPMLRRTSAQOGPSRGTSAFGALHGKKNSTYDCGV 600
601 VSLDAGPEATSPSHLLRPVMLBHPPTTTPSEPGGPQMLTQAPCVDSFEPPGARQ 660
601 VSLDAGPEATSPSHLLRPVMLBHPPTTTPSEPGGPQMLTQAPCVDSFEPPGARQ 660
661 RALSASVLTSLAELEESRHKCPCCMNLAQRYLITWECPLMMSIKQSVKLVMDPFTD 720
661 RALSASVLTSLAELEESRHKCPCCMNLAQRYLITWECPLMMSIKQSVKLVMDPFTD 720
721 LITWCIIVANTLPMALHNMTSEPEEMLOVGNLVFTGIFTAEMTKIILADPYYYFOG 780
721 LITWCIIVANTLPMALHNMTSEPEEMLOVGNLVFTGIFTAEMTKIILADPYYYFOG 780
781 WAFPSIIVITLSLMEGLSRMSNLSTLRSFRLLRPKAKSWPTNTLTKIIGNSVGALG 840
781 WAFPSIIVITLSLMEGLSRMSNLSTLRSFRLLRPKAKSWPTNTLTKIIGNSVGALG 840
841 NTLVLAIVFIFAVVGQLPFGKNSSELSDSGLLPRWHMDFFHAFILIRIILGEMI 900
841 NTLVLAIVFIFAVVGQLPFGKNSSELSDSGLLPRWHMDFFHAFILIRIILGEMI 900

901 ETMMDCHEVSGSGLCLLVFLVMVIGNLVYANFLALILSSFSADNLTAPEDEKEMNLQ 960
901 ETMMDCHEVSGSGLCLLVFLVMVIGNLVYANFLALILSSFSADNLTAPEDEKEMNLQ 960
961 LALARIORGRLFYKRTTWDFCCGLLRQRPQKAPALAAQGLPSCIATPYSPPEPEKVP 1020
961 LALARIORGRLFYKRTTWDFCCGLLRQRPQKAPALAAQGLPSCIATPYSPPEPEKVP 1020
1021 PTRKETFESEGPQCGTPEDEPVCYPIAVASDTDDGEDEENSLGTEESSKQESOP 1080
1021 PTRKETFESEGPQCGTPEDEPVCYPIAVASDTDDGEDEENSLGTEESSKQESOP 1080
1081 VSGPEAPPSKRTMSQVATASSEAEASQAQMRQMKAPAPGCGEPPESSCEBGT 1140
1081 VSGPEAPPSKRTMSQVATASSEAEASQAQMRQMKAPAPGCGEPPESSCEBGT 1140
1141 ADMNTAELEEQIPDLQDVKDEDCFTGECVRCPCCAVDTTQAPKQVMRLKTCYHI 1200
1141 ADMNTAELEEQIPDLQDVKDEDCFTGECVRCPCCAVDTTQAPKQVMRLKTCYHI 1200
1201 VEHSMFETPIIFMILSSGALAEEDYLERKTIKYLEADCMFTYVFLMMLKVMAY 1260
1201 VEHSMFETPIIFMILSSGALAEEDYLERKTIKYLEADCMFTYVFLMMLKVMAY 1260
1261 GFKKYFTNACWMLDPLVDVSVLVANTLGFAMGPIKSLRTLALRPLALSRFEGMR 1320
1261 GFKKYFTNACWMLDPLVDVSVLVANTLGFAMGPIKSLRTLALRPLALSRFEGMR 1320
1321 VVNVNALVGLPSIMNVLLVCLIFMLIFSINGVNLPAKFGRCINQTEGDIPLNTYIVNNK 1380
1321 VVNVNALVGLPSIMNVLLVCLIFMLIFSINGVNLPAKFGRCINQTEGDIPLNTYIVNNK 1380
1381 SOCESLNLTEGLWYTKKVNPDVNGAGYLLAQVATFKGMMDIMYAVDSRGVEEQWE 1440
1381 SOCESLNLTEGLWYTKKVNPDVNGAGYLLAQVATFKGMMDIMYAVDSRGVEEQWE 1440
1441 YNLVMTYFVFIPIFGSFFTLNLPFGVINDFNQOKKGLGODIEMTEBOKKYNAMKUL 1500
1441 YNLVMTYFVFIPIFGSFFTLNLPFGVINDFNQOKKGLGODIEMTEBOKKYNAMKUL 1500
1501 GSKKQKPIPRPLNKYOGFIIDYTKQAPDVITMFLCLNMVTMVTETDQSEPKINILA 1560
1501 GSKKQKPIPRPLNKYOGFIIDYTKQAPDVITMFLCLNMVTMVTETDQSEPKINILA 1560
1561 KINILFVAITGECIVKLAALRHYYFPNSWNIPFVVVLIISTGVTSIDIIQKTFSPTL 1620
1561 KINILFVAITGECIVKLAALRHYYFPNSWNIPFVVVLIISTGVTSIDIIQKTFSPTL 1620
1621 FFRVRLARIGRIILIRGAGIRITLFPALMMSLPALFNIGLLFLVMFIYSIFGMANFAY 1680
1621 FFRVRLARIGRIILIRGAGIRITLFPALMMSLPALFNIGLLFLVMFIYSIFGMANFAY 1680
1681 VKMEAGIDDMENFQTFANSMCLFOITTSAGMDLSPILINTGPYCDPLTPNSNGRGD 1740
1681 VKMEAGIDDMENFQTFANSMCLFOITTSAGMDLSPILINTGPYCDPLTPNSNGRGD 1740
1741 CGSPAVGILFFTTYIIISFLVNMVTAIILENSVATESTEBLSDDDMFEIEMKF 1800
1741 CGSPAVGILFFTTYIIISFLVNMVTAIILENSVATESTEBLSDDDMFEIEMKF 1800
1801 DPEATOFIEVSVDSPADALSEPIRIAKPNQISLIINDLPMVSGDRJHCHMDILFAFTKRV 1860
1801 DPEATOFIEVSVDSPADALSEPIRIAKPNQISLIINDLPMVSGDRJHCHMDILFAFTKRV 1860
1861 LGESGEMDALKIOMEKFMANPSKISYEPIITTLRKHEEVSAMVIQAFRRHLQSL 1920
1861 LGESGEMDALKIOMEKFMANPSKISYEPIITTLRKHEEVSAMVIQAFRRHLQSL 1920
1921 KHASFLPROAGSGLSBEDABERGLIAYVMSSEFSPRLGPPSSSISSTSPSPSYSVT 1980
1921 KHASFLPROAGSGLSBEDABERGLIAYVMSSEFSPRLGPPSSSISSTSPSPSYSVT 1980
1921 KHASFLPROAGSGLSBEDABERGLIAYVMSSEFSPRLGPPSSSISSTSPSPSYSVT 1980

QY 1981 RATSNDLQVRGSDYSHSEDLADPPSPDRRESIV 2015
 DB 1981 RATSNDLQVRGSDYSHSEDLADPPSPDRRESIV 2015

RESULT 4
 AEB22972
 ID AEB22972 standard; protein; 2015 AA.
 XX
 AC AEB22972;
 XX
 DT 22-SEP-2005 (first entry)
 XX
 DE Sodium channel alpha subunit human SCN5A gene derived protein.
 XX
 KW SCN5A; sodium channel; Brugada syndrome; heart arrhythmia;
 XX antiarrhythmic; selectable marker; ventricular fibrillation.
 OS Homo sapiens.
 XX JP005192413-A.
 XX
 XX PD 21-JUL-2005.
 XX
 XX PF 26-DEC-2003; 2003JP-00435234.
 XX
 XX PR 26-DEC-2003; 2003JP-00435234.
 XX
 XX (DOKU-) DOKURITSU GYOSEI HOJIN KAGAKU GIUTSU SH.
 PA
 PI Matenaga A;
 XX
 XX PI WPI; 2005-501993/51.
 DR
 XX Novel SCN5A gene of sodium channel alpha subunit, with mutations in which
 PT glycine being substituted by serine, and serine being substituted by
 PT leucine, at specific positions, useful as marker for diagnosing Brugada
 PT syndrome.
 XX
 PS Disclosure; SEQ ID NO 1; 30pp; Japanese.
 XX
 CC The invention relates to an novel SCN5A gene of a sodium channel alpha
 CC subunit. The novel SCN5A gene comprises the mutation G292S between the
 CC fifth and sixth membrane passing-through subunits of the first domain;
 CC and/or the mutation S835L of the intracellular loop between the fourth
 CC and fifth membrane passing-through subunits of the second domain. The
 CC novel SCN5A gene can be used as a Brugada syndrome marker, for diagnosing
 CC Brugada syndrome. Brugada syndrome causes idiopathic ventricular
 CC fibrillation, thus the gene/marker allow for the prevention or treatment
 CC of the syndrome. This sequence represents the protein of the wild-type
 CC human SCN5A gene of the invention.
 CC
 CC Sequence 2015 AA;
 SQ

Query Match 99.9%; Score 10484; DB 9; Length 2015;
 Best Local Similarity 99.9%; Pred. No. 0;
 Matches 2014; Conservative 0; Mismatches 1; Indels 0; Gaps 0;

QY 1 MANFLPRGTSFRFTRESLAIAIKMAEKQARGSTTLQESREGLPREEAPRPOLDIOA 60
 DB 1 MANFLPRGTSFRFTRESLAIAIKMAEKQARGSTTLQESREGLPREEAPRPOLDIOA 60
 QY 61 SKGLPDLGNPQELIGEPLEDDLPFYSTOKTFYIANKGTFIRPSATNALVYLSPPHPI 120
 DB 61 SKGLPDLGNPQELIGEPLEDDLPFYSTOKTFYIANKGTFIRPSATNALVYLSPPHPI 120
 QY 121 RRAAVKIIVHSLFNNLIMCTITLNCVFMAOHDPPEWTKYVEYTFATITPESLVKILARG 180
 DB 121 RRAAVKIIVHSLFNNLIMCTITLNCVFMAOHDPPEWTKYVEYTFATITPESLVKILARG 180
 QY 181 FCLHAFTELDPFWMNLDPDSVIIIMAYTTEFVDLGNVSALRTRFVRLALXTISVISGLKTIIV 240
 DB 181 FCLHAFTELDPFWMNLDPDSVIIIMAYTTEFVDLGNVSALRTRFVRLALXTISVISGLKTIIV 240

QY 241 GALIOSYKKGADAVWVLTJFVCLSVFALIGLQFMGNLRHKCVNFTALANGTNGSVBADGLV 300
 DB 241 GALIOSYKKGADAVWVLTJFVCLSVFALIGLQFMGNLRHKCVNFTALANGTNGSVBADGLV 300
 QY 301 WESLDLYLSDPENYLLKNGTSDVLLCGNSSDAGTCPEGYRCUKAGBNPDHGYTSFDSFAM 360
 DB 301 WESLDLYLSDPENYLLKNGTSDVLLCGNSSDAGTCPEGYRCUKAGBNPDHGYTSFDSFAM 360
 QY 361 AFLALFRLMTQDCWRLYQOTLRAGKTYMIFFMVLVILGSPYLVNLIIVAVAMAYEON 420
 DB 361 AFLALFRLMTQDCWRLYQOTLRAGKTYMIFFMVLVILGSPYLVNLIIVAVAMAYEON 420
 QY 421 QATIAETEKEKRPQEAEMMLKKEHEALTIRGVDTVSRSLSLMSPLAPNHSERSKRRK 480
 DB 421 QATIAETEKEKRPQEAEMMLKKEHEALTIRGVDTVSRSLSLMSPLAPNHSERSKRRK 480
 QY 481 RMSSGTEECGEDRLPKSDSEDPGRAMNHLSTRGLSRTSMKPRSSRGSIFFRRRDLGSE 540
 DB 481 RMSSGTEECGEDRLPKSDSEDPGRAMNHLSTRGLSRTSMKPRSSRGSIFFRRRDLGSE 540
 QY 541 ADFADDENSTAGESSHRTSLVWPILRRTSAQCGPSPGTSAPGHALHGXNXTVDGCV 600
 DB 541 ADFADDENSTAGESSHRTSLVWPILRRTSAQCGPSPGTSAPGHALHGXNXTVDGCV 600
 QY 601 VSLDAGDPEATSPGSHLLRPVMLEHPDPTTPSSEBPQOMLTQACVCGFEERGARQ 660
 DB 601 VSLDAGDPEATSPGSHLLRPVMLEHPDPTTPSSEBPQOMLTQACVCGFEERGARQ 660
 QY 661 RALSASVYLTSALIEESRHKCPQCNRLAQRVLIWECCLPMWSIKOGVKLVMDPFTD 720
 DB 661 RALSASVYLTSALIEESRHKCPQCNRLAQRVLIWECCLPMWSIKOGVKLVMDPFTD 720
 QY 721 LTTWCIVLNTLFMALHYNMTSEFEEMLOQGNLVFTGIFLAEMTFKIIALDPYYTFOQG 780
 DB 721 LTTWCIVLNTLFMALHYNMTSEFEEMLOQGNLVFTGIFLAEMTFKIIALDPYYTFOQG 780
 QY 781 WNIPISTIIVILSIELGSRMSNLVSRSPFLLRPKAKSMPTNTLIIKIIIGNSVGALG 840
 DB 781 WNIPISTIIVILSIELGSRMSNLVSRSPFLLRPKAKSMPTNTLIIKIIIGNSVGALG 840
 QY 841 NLTLVIAITVFIFAVGQGLFGKNYSLELRDSDGLLPRMHMDFFHAFLIIFRILGEMI 900
 DB 841 NLTLVIAITVFIFAVGQGLFGKNYSLELRDSDGLLPRMHMDFFHAFLIIFRILGEMI 900
 QY 901 ETMDMCEVSGQSCLVFLVWYIGNLVNLFALILSSFSADNLTAPDEDRMNNLIQ 960
 DB 901 ETMDMCEVSGQSCLVFLVWYIGNLVNLFALILSSFSADNLTAPDEDRMNNLIQ 960
 QY 961 LALARIQGLRFVYKRTTWDGCCGLLRQRPQAPALAAQOLPSCITATYSPPPETEKYP 1020
 DB 961 LALARIQGLRFVYKRTTWDGCCGLLRQRPQAPALAAQOLPSCITATYSPPPETEKYP 1020
 QY 1021 PTRKRETRBEEOGQGTGPDPPEPCVPIAAESPTDQOEDEENSLTEBESSSQOESOP 1080
 DB 1021 PTRKRETRBEEOGQGTGPDPPEPCVPIAAESPTDQOEDEENSLTEBESSSQOESOP 1080
 QY 1081 VSGGPEAPDPSRTSVOVSATASSEAEASASQADWRQKAPQAPGCGETPDSSEBST 1140
 DB 1081 VSGGPEAPDPSRTSVOVSATASSEAEASASQADWRQKAPQAPGCGETPDSSEBST 1140
 QY 1141 ADMNTNIALEQIPDLGQDVDPEDCFTEGCVRCPCCAVDTTQAPGVMWRRLKRTCYHI 1200
 DB 1141 ADMNTNIALEQIPDLGQDVDPEDCFTEGCVRCPCCAVDTTQAPGVMWRRLKRTCYHI 1200
 QY 1201 VEHSMFETFIIFMILSSGALAFEDIVLEBKRTIKVLEIYADKQFTYFVLEMLIKWAY 1260
 DB 1201 VEHSMFETFIIFMILSSGALAFEDIVLEBKRTIKVLEIYADKQFTYFVLEMLIKWAY 1260
 QY 1261 GPKKYFTNAMCMLDFLIYDVSLSLVANTLGFAEMGPIKSLRTLRALRPLALSREFGMR 1320
 DB 1261 GPKKYFTNAMCMLDFLIYDVSLSLVANTLGFAEMGPIKSLRTLRALRPLALSREFGMR 1320

Qy	1321	VVNVALVGAIPSIANNVLVCLIPILISINGVNLFAKPGACINOTEGDLPYNTYIYNNK	1380
Db	1321	VVNVALVGAIPSIANNVLVCLIPILISINGVNLFAKPGACINOTEGDLPYNTYIYNNK	1380
Qy	1381	SOCESLNLZBELYWTKVKNFNDVNGAGYLLAQVATFKGMDIMYAADSRGIEBOPQME	1440
Db	1381	SOCESLNLZBELYWTKVKNFNDVNGAGYLLAQVATFKGMDIMYAADSRGIEBOPQME	1440
Qy	1441	YNLYMYTYVYFIIRGSSPPTLNLFIGYIDNPNQKKKLGGODIFMTBEOKKYNNAKKL	1500
Db	1441	YNLYMYTYVYFIIRGSSPPTLNLFIGYIDNPNQKKKLGGODIFMTBEOKKYNNAKKL	1500
Qy	1501	GSKKPQKPIRPLNKYOGFIEDIYTKOAFDVTIMFLI CLANNVTMVEITDQSPKINILA	1560
Db	1501	GSKKPQKPIRPLNKYOGFIEDIYTKOAFDVTIMFLI CLANNVTMVEITDQSPKINILA	1560
Qy	1561	KINLLPVALPFGECIVLALALRHYHFNPSNMKIPEPVVILISIVGTULSDIIOKXFSPPTL	1620
Db	1561	KINLLPVALPFGECIVLALALRHYHFNPSNMKIPEPVVILISIVGTULSDIIOKXFSPPTL	1620
Qy	1621	FRVIRLARIGIIRLIRIGAKGIRFLLPALMMSLPALFNIGLLLVNFYISIFGMANFAY	1680
Db	1621	FRVIRLARIGIIRLIRIGAKGIRFLLPALMMSLPALFNIGLLLVNFYISIFGMANFAY	1680
Qy	1681	VKMEAGIDDMNFOTFANSMULCFQIITTSAGMDGLSPILANTGPYCDPTLPNSNGSRGD	1740
Db	1681	VKMEAGIDDMNFOTFANSMULCFQIITTSAGMDGLSPILANTGPYCDPTLPNSNGSRGD	1740
Qy	1741	CGSPAVGILFPTTYIIISFLIVANNMYAILIENFSVATEESTEPLESDPFMFIEIWEKF	1800
Db	1741	CGSPAVGILFPTTYIIISFLIVANNMYAILIENFSVATEESTEPLESDPFMFIEIWEKF	1800
Qy	1801	DPEATOFIERYSLSDPADALSEPLRIAKPNOISILINDLPMVSGGRHCHMDILPAFTKRV	1860
Db	1801	DPEATOFIERYSLSDPADALSEPLRIAKPNOISILINDLPMVSGGRHCHMDILPAFTKRV	1860
Qy	1861	LGSEGGENDALKIOMEKEMFMANPXSISYEPITTLTLRRGHEVSAWVIOARFRRLHQRSL	1920
Db	1861	LGSEGGENDALKIOMEKEMFMANPXSISYEPITTLTLRRGHEVSAWVIOARFRRLHQRSL	1920
Qy	1921	KHASFLEFROQAGSGISEDAPEREGLIAYVNSENFSRPLGPPSSSISISTSPPSYDSVT	1980
Db	1921	KHASFLEFROQAGSGISEDAPEREGLIAYVNSENFSRPLGPPSSSISISTSPPSYDSVT	1980
Qy	1981	RATSDNLOVRSGDYSHSEDLADFPSPBPRDEBSIV 2015	
Db	1981	RATSDNLOVRSGDYSHSEDLADFPSPBPRDEBSIV 2015	

RESULT 5	
ADW33997	
ID	ADW33997 standard; protein; 2016 AA.
XX	
AC	ADW33997;
XX	
DT	03-JUN-2004 (first entry)
XX	
DE	Human SCN5A variant 2 protein SEQ ID NO:4.
XX	
KW	human; cardiac sodium channel alpha subunit; SCN5A;
KW	sodium voltage-gated channel; type V; alpha; mutation study;
KW	sodium channel related disease.
XX	
OS	Homo sapiens.
XX	
PN	WO2004012668-A2.
XX	
PD	12-FEB-2004.
XX	
PF	01-AUG-2003; 2003WO-US024190.
XX	
PR	02-AUG-2002; 2002US-040101BP.
XX	

PA (WISC) WISCONSIN ALUMNI RES FOUND.
XX
XX
PI Makielski JC, Ye B, Ackerman MJ;
XX
XX WPI; 2004-157000/15.
DR N-PSDB; ADM33996.
XX
XX
XX New nucleic acid encoding sodium voltage-gated channel, type V, alpha
PT polypeptide variants, useful for studying mutations, and designing or
PT identifying new diagnostics and treatment strategies or agents for sodium
PT channel related diseases.
XX
XX
PS Claim 11; SEQ ID NO 4; 11pp; English.

The present sequence represents a variant of the human cardiac sodium channel alpha subunit designated SCN5A (sodium voltage-gated channel, type V, alpha) protein. The present invention describes an isolated polynucleotide encoding an SCN5A polypeptide where the polynucleotide is selected from the group: (1) a first polynucleotide that encodes an SCN5A polypeptide selected from the group consisting of: (i) a histidine, threonine, leucine, arginine and glutamine at amino acid positions 558, 559, 618, 1027 and 1077, respectively; (ii) an arginine, threonine, leucine, arginine and glutamine at amino acid positions 558, 559, 618, 1027 and 1077, respectively; (iii) a histidine, threonine, leucine and arginine at amino acid positions 558, 559, 618 and 1027, respectively, with the amino acid at amino acid position 1077 deleted; or (iv) an arginine, threonine, leucine and arginine at amino acid positions 558, 559, 618 and 1027, respectively, with the amino acid at amino acid position 1077 deleted; (2) a second polynucleotide that is at least 80 % identical to the first polynucleotide over the entire length of the first polypeptide; (3) a third polynucleotide that encodes any of the SCN5A polypeptides with a conservative substitution, deletion or rearrangement at one or more non-critical amino acid position; and (4) a fourth polynucleotide that is a complement of the first, second or third polynucleotide. Also described: (1) a genetic construct comprising the polynucleotide operably linked to a non-native expression control sequence; (2) a cell comprising the polynucleotide; (3) an isolated polypeptide encoded by the polynucleotide; (4) an antibody that specifically binds to the polypeptide; (5) identifying an agent that can alter the expression of a sodium channel; (6) identifying an agent that can alter the expression of a sodium channel; (7) determining whether a biological sample or a preparation derived from the biological sample contains the polypeptide; (8) determining whether a mutation on a sodium channel is associated with a disease; and (9) determining whether a human or non-human subject is at risk for Long QT syndrome. The SCN5A polynucleotides and polypeptides are useful for studying mutations, and designing or identifying new diagnostics and treatment strategies or agents for sodium channel related diseases or conditions.

Sequence 2016 AA;

Query Match	99.9%	Score 10478.5	DB 8	Length 2016
Best Local Similarity	99.9%	Pred. No. 0		
Matches 2015; Conservative	0	Mismatches	0	Indels 1; Gaps 1

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QY      1  MANFLPBGTSFREFTEESLAAEKMAEKOARSTLQSSRELPREEAPRPLDLOQA  60
Db      1  MANFLPBGTSFREFTEESLAAEKMAEKOARSTLQSSRELPREEAPRPLDLOQA  60
QY      61  SKCLPDLYGNPPQELIGEBLEDDBPFYSTOKTFYLANGKTIIPRSANNAIYLSPPHPI  120
Db      61  SKCLPDLYGNPPQELIGEBLEDDBPFYSTOKTFYLANGKTIIPRSANNAIYLSPPHPI  120
QY      121  RRAAVKIIIVSHLFNNLIMCTIITLNCVMAOHDPPWTKYVEYTFATITYESLVKILARG  180
Db      121  RRAAVKIIIVSHLFNNLIMCTIITLNCVMAOHDPPWTKYVEYTFATITYESLVKILARG  180
QY      181  PCLHAFPTLRDPMNLDFSVIIIMAYTTEFVDLGNVSAIIRTFRVALAKTISVISGLKTIY  240
Db      181  PCLHAFPTLRDPMNLDFSVIIIMAYTTEFVDLGNVSAIIRTFRVALAKTISVISGLKTIY  240
QY      241  TALLISVKKLADVMVLTYFCLSVFALLIGOLFPMGLRHRKCVNFIALNGTSSVADGIV  300

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Db 241 GALLIOSVKKLADVWMLTVFCLSVFALIGLQFMWGNLRHKCVENFTALNTNGSVSEADGLV 300
 QY 301 WESLILYSDPENNYLLKNGTSDVLLCGNSSDAGTCEGRCLKAGENPHGTSPPSFAW 360
 Db 301 WESLILYSDPENNYLLKNGTSDVLLCGNSSDAGTCEGRCLKAGENPHGTSPPSFAW 360
 QY 361 AFLALFRLMTQOCWERLYOQTLSRAGKIYIMFFMLYIFLGSFYLVLVLAVAMAYEON 420
 Db 361 AFLALFRLMTQOCWERLYOQTLSRAGKIYIMFFMLYIFLGSFYLVLVLAVAMAYEON 420
 QY 421 QATIAETEBEKRPOEAMEMLKKEHEALTRGVDTVSRSLSLMSPLAPVNSHERSKRK 480
 Db 421 QATIAETEBEKRPOEAMEMLKKEHEALTRGVDTVSRSLSLMSPLAPVNSHERSKRK 480
 QY 481 RUSSTCEGCEGRLLPSSDEDEPRANMHLSTRGLSRTSMKPRSSGSIPTRRRLDGE 540
 Db 481 RUSSTCEGCEGRLLPSSDEDEPRANMHLSTRGLSRTSMKPRSSGSIPTRRRLDGE 540
 QY 541 ADPADENSTAGESESHRTSLVPMPLRRTSAQOGSPGTSAPGHALHKKNSYVDCNV 600
 Db 541 ADPADENSTAGESESHRTSLVPMPLRRTSAQOGSPGTSAPGHALHKKNSYVDCNV 600
 QY 601 VSLLAGDPEATSPGSHLLRPVMLEBHPDPTTPEBPGGPOMLTSQAPCVDFEERGAQ 660
 Db 601 VSLLAGDPEATSPGSHLLRPVMLEBHPDPTTPEBPGGPOMLTSQAPCVDFEERGAQ 660
 QY 661 RALSASVUTSLALBEEBRRHKCPGCMNLAORYLIMECCPLMMSIKQYKAVMDPFD 720
 Db 661 RALSASVUTSLALBEEBRRHKCPGCMNLAORYLIMECCPLMMSIKQYKAVMDPFD 720
 QY 721 LITIMCIYANTLPMALBHNMTSBPEBMLQVGNLVTGIFTAEMTKIALDPYFYFOG 780
 Db 721 LITIMCIYANTLPMALBHNMTSBPEBMLQVGNLVTGIFTAEMTKIALDPYFYFOG 780
 QY 781 WNIPIISIIYLSLMEIGLSRMSNLSTYRSFRLRYFKLAKSWPTLNTLKIIGNSYGALG 840
 Db 781 WNIPIISIIYLSLMEIGLSRMSNLSTYRSFRLRYFKLAKSWPTLNTLKIIGNSYGALG 840
 QY 841 NITLVLAIVTIPAVYGMQLFGKNTSELDSGSLPRWMDFFRAFLITRIICGEVI 900
 Db 841 NITLVLAIVTIPAVYGMQLFGKNTSELDSGSLPRWMDFFRAFLITRIICGEVI 900
 QY 901 ETTMDCMEVSGOSCLVFLVLMVIGNLVNLFLALILSSPSADNLTPADEDRMNNIQ 960
 Db 901 ETTMDCMEVSGOSCLVFLVLMVIGNLVNLFLALILSSPSADNLTPADEDRMNNIQ 960
 QY 961 LALARIORGARFVKETWDFCCGLRORPOKPAALAAQOLPSCIATPYSPPETEKVP 1020
 Db 961 LALARIORGARFVKETWDFCCGLRORPOKPAALAAQOLPSCIATPYSPPETEKVP 1020
 QY 1021 PTRKETREBEGHQPQGTIPGDEPVCPIAIAESDTDOEDBENS LGTEBESSK_QESQ 1079
 Db 1021 PTRKETREBEGHQPQGTIPGDEPVCPIAIAESDTDOEDBENS LGTEBESSK_QESQ 1079
 QY 1080 PVSGBPEAPDPSRTMSOVSATASSSEAEASQAADRQOKAPAPQGCETEDSCSBS 1139
 Db 1080 PVSGBPEAPDPSRTMSOVSATASSSEAEASQAADRQOKAPAPQGCETEDSCSBS 1139
 QY 1140 TADMTNTAELLEQIDLDGQVNDPEDCFTEGCVRRCPCCAVDTTQAPGVMMRLKTCYH 1199
 Db 1140 TADMTNTAELLEQIDLDGQVNDPEDCFTEGCVRRCPCCAVDTTQAPGVMMRLKTCYH 1199
 QY 1200 IYHSWSEFTFIIIFMILLSSGALAFEDYIEBKRTIKVLEVDKMTTYFVLEMLIKMYA 1259
 Db 1200 IYHSWSEFTFIIIFMILLSSGALAFEDYIEBKRTIKVLEVDKMTTYFVLEMLIKMYA 1259
 QY 1260 YGFKKYPFNAMCMLDFLIVDSVLVSVANTLGPAMGPIKSLRTLRALRPLALSRFEGM 1319
 Db 1260 YGFKKYPFNAMCMLDFLIVDSVLVSVANTLGPAMGPIKSLRTLRALRPLALSRFEGM 1319
 QY 1320 RVVVALVGAISIMNVLLVCLIFMLIFSIMGVNLFAGKFGRCINOTEGDLPLANTYVNN 1379
 Db 1320 RVVVALVGAISIMNVLLVCLIFMLIFSIMGVNLFAGKFGRCINOTEGDLPLANTYVNN 1379
 QY 1321 RVVVALVGAISIMNVLLVCLIFMLIFSIMGVNLFAGKFGRCINOTEGDLPLANTYVNN 1380
 Db 1321 RVVVALVGAISIMNVLLVCLIFMLIFSIMGVNLFAGKFGRCINOTEGDLPLANTYVNN 1380

QY 1380 KSQCESLNLVTGELYMTKVKVNFNDVAGAGYIALLOVATFKGMDIMYAADVSRGYEQPOM 1439
 Db 1381 KSQCESLNLVTGELYMTKVKVNFNDVAGAGYIALLOVATFKGMDIMYAADVSRGYEQPOM 1440
 QY 1440 EYNLYMYIYFVFIIFGSEFTLNLFIYIINDFNQOKKKGGQDIFMTEBOKKYVAMAK 1499
 Db 1441 EYNLYMYIYFVFIIFGSEFTLNLFIYIINDFNQOKKKGGQDIFMTEBOKKYVAMAK 1500
 QY 1500 LGSKKPQKPIPRPLANKYGFIDYITKQAFDYTIMFLICLANNVTMMVETDDOSPEKINIL 1559
 Db 1501 LGSKKPQKPIPRPLANKYGFIDYITKQAFDYTIMFLICLANNVTMMVETDDOSPEKINIL 1560
 QY 1560 AKINILFAIFNGECIVKLAALRHYFFNNSWNIIPFVVYIISVGTVLSDIIOKTFEFT 1619
 Db 1561 AKINILFAIFNGECIVKLAALRHYFFNNSWNIIPFVVYIISVGTVLSDIIOKTFEFT 1620
 QY 1620 LFRVIRLARIGRILRLIRGAKGIRTLFALMMSLPALFNIGLLFLVMTYSIFGMANPA 1679
 Db 1621 LFRVIRLARIGRILRLIRGAKGIRTLFALMMSLPALFNIGLLFLVMTYSIFGMANPA 1680
 QY 1680 YKMEAGIDDMNFOTFANSMLCLFOITTSAGWDGLSPILNTGSPYCDPLPNSNGSRG 1739
 Db 1681 YKMEAGIDDMNFOTFANSMLCLFOITTSAGWDGLSPILNTGSPYCDPLPNSNGSRG 1740
 QY 1740 DCGSPAVGILFETTYIIISPLIVNMVYIAIILENFSVATESSTEBLSBDPMPYEIWEK 1799
 Db 1741 DCGSPAVGILFETTYIIISPLIVNMVYIAIILENFSVATESSTEBLSBDPMPYEIWEK 1800
 QY 1800 FDPPEATOPTEYSVLSDFADALSEPRLIAKPNQISLIMNDLPVSGDRHICMDILFAFTR 1859
 Db 1801 FDPPEATOPTEYSVLSDFADALSEPRLIAKPNQISLIMNDLPVSGDRHICMDILFAFTR 1860
 QY 1860 VLGSEGDMDALKIOMEKFMANPSKISYEPIITTLRRKHEVSAMVIOARFRHLORS 1919
 Db 1861 VLGSEGDMDALKIOMEKFMANPSKISYEPIITTLRRKHEVSAMVIOARFRHLORS 1920
 QY 1920 LKHSFLLPQOAGSLSEBDAPERGLIAYVMSNPSRPLGPPSSSISSTSPSPSYDSV 1979
 Db 1921 LKHSFLLPQOAGSLSEBDAPERGLIAYVMSNPSRPLGPPSSSISSTSPSPSYDSV 1980
 QY 1980 TRATSDNLQVRSQVSHSEDLADPPSPDRDRESIV 2015
 Db 1981 TRATSDNLQVRSQVSHSEDLADPPSPDRDRESIV 2016

RESULT 6
 AEB22993
 ID AEB22993 standard; protein; 2015 AA.
 XX
 AC AEB22993;
 XX
 AC 22-SBP-2005 (first entry)
 XX
 DE Mutant human SCNSA protein, S935L.
 XX
 KW SCNSA; sodium channel; Brugada syndrome; heart arrhythmia;
 XX antiarrhythmic; selectable marker; ventricular fibrillation; mutein.
 OS Homo sapiens.
 XX
 FT Key Location/Qualifiers
 FT Misc-difference 835
 FT FT /note= "The wild-type Ser residue has been substituted
 FT with Leu"
 XX
 PN JP2005192413-A.
 XX
 PD 21-JUL-2005.
 XX
 PF 26-DEC-2003; 2003JP-00435234.
 XX

PR 26-DEC-2003; 2003JF-00435234.
XX (DOKU-) DOKURITSU GYOSHI HOJIN KAGAKU GIJUTSU SH.
XX Matsunaga A;
PI WPI; 2005-501993/51.
DR
XX Novel SCN5A gene of sodium channel alpha subunit, with mutations in which
PT glycine being substituted by serine, and serine being substituted by
PT leucine, at specific positions, useful as marker for diagnosing Brugada
PT syndrome.
XX
PS Claim 1; Page: 30pp; Japanese.
XX
XX The invention relates to an novel SCN5A gene of a sodium channel alpha
CC subunit. The novel SCN5A gene comprises the mutation G292S between the
CC fifth and sixth membrane passing-through subunits of the first domain;
CC and/or the mutation S835L of the intracellular loop between the fourth
CC and fifth membrane passing-through subunits of the second domain. The
CC novel SCN5A gene can be used as a Brugada syndrome marker, for diagnosing
CC Brugada syndrome. Brugada syndrome causes idiopathic ventricular
CC fibrillation, thus the gene/marker allow for the prevention or treatment
CC of the syndrome. This sequence represents the S835L mutant protein of the
CC human SCN5A gene of the invention.
XX
SQ Sequence 2015 AA;
Query Match 99.9%; Score 10478; DB 9; Length 2015;
Best Local Similarity 99.9%; Pred. No. 0;
Matches 2013; Conservative 0; Mismatches 2; Indels 0; Gaps 0;
QY 1 MANFLPFGTSSFRFTBESLAIRKMAEKQARSTLQESREGLPREEAPRPDLQA 60
DB 1 MANFLPFGTSSFRFTBESLAIRKMAEKQARSTLQESREGLPREEAPRPDLQA 60
QY 61 SKGLPDLVGNPPOELIGEPLEDDLPFYSTQKTFIYLNKGTIFRSATNALVLSPPFI 120
DB 61 SKGLPDLVGNPPOELIGEPLEDDLPFYSTQKTFIYLNKGTIFRSATNALVLSPPFI 120
QY 121 RRAAVKIIVHSLFNNMLIMCTILTNCFVMAQHPPMWTKVXYETFTAIYFESLVKILAR 180
DB 121 RRAAVKIIVHSLFNNMLIMCTILTNCFVMAQHPPMWTKVXYETFTAIYFESLVKILAR 180
QY 181 FCLAAFTFLRDPWNNLDSVITIMAYTTEFVDLGNVSAIRTPRVBALKTIVISGLKTIY 240
DB 181 FCLAAFTFLRDPWNNLDSVITIMAYTTEFVDLGNVSAIRTPRVBALKTIVISGLKTIY 240
QY 241 FCLAAFTFLRDPWNNLDSVITIMAYTTEFVDLGNVSAIRTPRVBALKTIVISGLKTIY 240
DB 241 FCLAAFTFLRDPWNNLDSVITIMAYTTEFVDLGNVSAIRTPRVBALKTIVISGLKTIY 240
QY 241 GALISVKKLADVWVLTVCISVPALIGQLFMGNLRHKCVNFTALNGTNGSVBADGLV 300
DB 241 GALISVKKLADVWVLTVCISVPALIGQLFMGNLRHKCVNFTALNGTNGSVBADGLV 300
QY 301 WESLDLYLSDPENNYLLKNGTSDVLLCGNSSDAGTCEGYRCLKAGENPDHGYTSFDSFAM 360
DB 301 WESLDLYLSDPENNYLLKNGTSDVLLCGNSSDAGTCEGYRCLKAGENPDHGYTSFDSFAM 360
QY 361 AFLAFLRLMTODCWERLYOQTRSAGKIYIMFPMVIFIGSFYLVNLLAIVAMAYEEN 420
DB 361 AFLAFLRLMTODCWERLYOQTRSAGKIYIMFPMVIFIGSFYLVNLLAIVAMAYEEN 420
QY 421 QATIAETEERKEKRFQAMEMLKKEHEALITRGVDYTSRSSLEMSPLAPNHSERHSKRRK 480
DB 421 QATIAETEERKEKRFQAMEMLKKEHEALITRGVDYTSRSSLEMSPLAPNHSERHSKRRK 480
QY 481 RNSSGTEECGEDRLPKSDESDGPRANMHLSTRGLSRTSMKPRSSRGSIFTRRRDLGSE 540
DB 481 RNSSGTEECGEDRLPKSDESDGPRANMHLSTRGLSRTSMKPRSSRGSIFTRRRDLGSE 540
QY 541 ADPADDENSTAGESSEHRTSLVWPVLRRTSAQGQSPGTSAPGALHKKNSYDPCNV 600
DB 541 ADPADDENSTAGESSEHRTSLVWPVLRRTSAQGQSPGTSAPGALHKKNSYDPCNV 600
QY 601 VSLLAGDPEATSPGSHLLRPVWLBNRPPTTTPSEBPGPOMLTSGAPCVDGFEERGARQ 660

DB VSLLAGDPEATSPGSHLLRPVWLBNRPPTTTPSEBPGPOMLTSGAPCVDGFEERGARQ 660
QY 661 RALSAYSVLTSALBEELSESRHKCPCCNRLAQRVYLMECCPLMWSIKQGVLYVMDPFTD 720
DB 661 RALSAYSVLTSALBEELSESRHKCPCCNRLAQRVYLMECCPLMWSIKQGVLYVMDPFTD 720
QY 721 LITTMCIIVNTLPMALHVNMTSEFERMLQVGNLVFTGIFTAEMTEKXIALDPPYFQOG 780
DB 721 LITTMCIIVNTLPMALHVNMTSEFERMLQVGNLVFTGIFTAEMTEKXIALDPPYFQOG 780
QY 781 WNIIFDSIIIVLSLMEIGLSMWSNLSTVRSRLLRVFKLASWPTLNTLKIIGNSVAGL 840
DB 781 WNIIFDSIIIVLSLMEIGLSMWSNLSTVRSRLLRVFKLASWPTLNTLKIIGNSVAGL 840
QY 841 NLTVLAIIVFIPAVNMOLFKNYSRLRSDSGLLPRHMDPFHAFILIFRILGEMI 900
DB 841 NLTVLAIIVFIPAVNMOLFKNYSRLRSDSGLLPRHMDPFHAFILIFRILGEMI 900
QY 901 ETMMDCEVSGOSICLLVFLVNVYGNLVNLFLALLSSFSADNLTADEDEKEMNLQ 960
DB 901 ETMMDCEVSGOSICLLVFLVNVYGNLVNLFLALLSSFSADNLTADEDEKEMNLQ 960
QY 961 LALARIQGLRFPVKRTTWDPCGILLRQRPKPAALAAQGLPSCIATPYSPPEETKVP 1020
DB 961 LALARIQGLRFPVKRTTWDPCGILLRQRPKPAALAAQGLPSCIATPYSPPEETKVP 1020
QY 1021 PTRKTEPREEGEOGCTPGDPPRVCPIVAVSDTDQDEDEENSLGTEESKQESOP 1080
DB 1021 PTRKTEPREEGEOGCTPGDPPRVCPIVAVSDTDQDEDEENSLGTEESKQESOP 1080
QY 1081 VSGGEAPRPSRTMSQVSATASSAEASASQADWRQWKAEPQAPGCGETPEDSCSEGST 1140
DB 1081 VSGGEAPRPSRTMSQVSATASSAEASASQADWRQWKAEPQAPGCGETPEDSCSEGST 1140
QY 1141 ADMNTAELLBOIPDLQDVDPEDCTBEGCVRCPCAVDTTQAPKQWWRILKTYHI 1200
DB 1141 ADMNTAELLBOIPDLQDVDPEDCTBEGCVRCPCAVDTTQAPKQWWRILKTYHI 1200
QY 1201 VEHSMPTFTIIFMLLSSGALAFEDYLBEEKITVLEBZADKMTFVFLMLKKVAY 1260
DB 1201 VEHSMPTFTIIFMLLSSGALAFEDYLBEEKITVLEBZADKMTFVFLMLKKVAY 1260
QY 1261 GFKKYFTNAMCMLDFLIVDSVLSLVANTLGAEMGPIKSLRTLALRPLRALSREFGMR 1320
DB 1261 GFKKYFTNAMCMLDFLIVDSVLSLVANTLGAEMGPIKSLRTLALRPLRALSREFGMR 1320
QY 1321 VVNAALVGAIPSIINVLVCLIFMLIFSINGVNI.PACKFGRCINOTEGDPLANTTYNNK 1380
DB 1321 VVNAALVGAIPSIINVLVCLIFMLIFSINGVNI.PACKFGRCINOTEGDPLANTTYNNK 1380
QY 1381 SCSLSLNTLGBLWTKVKNVDNAGAGYLLAQVATFKGMWDIYAAVDSRGYEQOME 1440
DB 1381 SCSLSLNTLGBLWTKVKNVDNAGAGYLLAQVATFKGMWDIYAAVDSRGYEQOME 1440
QY 1441 YNLWYIYFVFIIFGSEFTLNLFIYIINDFNQKKLGGQDI.FMTEBQKKYNNAMKL 1500
DB 1441 YNLWYIYFVFIIFGSEFTLNLFIYIINDFNQKKLGGQDI.FMTEBQKKYNNAMKL 1500
QY 1501 GSKKQKPIPRPLNKYQGFIDYITKQAFDVTIMFLCLNNVTMMWETDDOSPKNILA 1560
DB 1501 GSKKQKPIPRPLNKYQGFIDYITKQAFDVTIMFLCLNNVTMMWETDDOSPKNILA 1560
QY 1561 KINLLFVAIFPGECIVKLAALRHHYFPMNSWNI.PPVVVYIISIVGTVSDII.QKFFSPTL 1620
DB 1561 KINLLFVAIFPGECIVKLAALRHHYFPMNSWNI.PPVVVYIISIVGTVSDII.QKFFSPTL 1620
QY 1621 FRVIRLAIIGILRLIRGAKGIRTLFLPAMSLPALFNIGLFLPVEYISIFGMANFAY 1680
DB 1621 FRVIRLAIIGILRLIRGAKGIRTLFLPAMSLPALFNIGLFLPVEYISIFGMANFAY 1680
QY 1681 VKWEAGIDMFNPOTFANSMLCLFOITTSAGWDGLSPIILNTGPPYCDPTLPNSGSGCD 1740

QY 961 LALARIQGLRFEVKTTHWDFCCGLLRORPOKPAALAAAGOLPSCIATPYSPPEETKVP 1020
 DB 961 LALARIQGLRFEVKTTHWDFCCGLLRORPOKPAALAAAGOLPSCIATPYSPPEETKVP 1020
 QY 1021 PTKRETRREEBEOQCGCTPGDPEPVCVPIAAVESPTDDOEDEENSLGTEEESSKOESOP 1080
 DB 1021 PTKRETRREEBEOQCGCTPGDPEPVCVPIAAVESPTDDOEDEENSLGTEEESSKOESOP 1080
 QY 1081 VSGPEAPDPSRTRYSQVATASSEAEASASQADMWQOMKAEPOAPGCEPTEDSCSEGST 1140
 DB 1081 VSGPEAPDPSRTRYSQVATASSEAEASASQADMWQOMKAEPOAPGCEPTEDSCSEGST 1140
 QY 1141 ADMNTAELFOIPLGQDVDPEDCFTEGCVRRCPCCAVDTQAPGVMWRIRKTCYHI 1200
 DB 1141 ADMNTAELFOIPLGQDVDPEDCFTEGCVRRCPCCAVDTQAPGVMWRIRKTCYHI 1200
 QY 1201 VEHSMFEFTIIFMILSSGALAFEDIYLEBKRTIKVLEBYADKMFTYFVLEMLKWAY 1260
 DB 1201 VEHSMFEFTIIFMILSSGALAFEDIYLEBKRTIKVLEBYADKMFTYFVLEMLKWAY 1260
 QY 1261 GPKKYFTINACWDLFLIVDSIVSVANTLQFAEMKPIKSLRTLALRPLALSRFEGMR 1320
 DB 1261 GPKKYFTINACWDLFLIVDSIVSVANTLQFAEMKPIKSLRTLALRPLALSRFEGMR 1320
 QY 1321 VVNAALVGAISIMWVLLVCLIFMLIFSIMGVNLFAGKRGRCINOTEGDLPLANTYVNNK 1380
 DB 1321 VVNAALVGAISIMWVLLVCLIFMLIFSIMGVNLFAGKRGRCINOTEGDLPLANTYVNNK 1380
 QY 1381 SQCESLANTGELWTKVKNFEDNVGAGYLAALQVATFEGMMDIMVAAVDSRGYEQPOME 1440
 DB 1381 SQCESLANTGELWTKVKNFEDNVGAGYLAALQVATFEGMMDIMVAAVDSRGYEQPOME 1440
 QY 1441 YNLWYIYFVPIIFGSEFTNLPLFVIIIDNFNOQKKLGGODIMTEBOKKYTNAMKUL 1500
 DB 1441 YNLWYIYFVPIIFGSEFTNLPLFVIIIDNFNOQKKLGGODIMTEBOKKYTNAMKUL 1500
 QY 1501 GSKKPKRPIPRPNKYOGPIPIDIVTKOAFDVTIMELICLNWTVWVEVDOSPEKINTILA 1560
 DB 1501 GSKKPKRPIPRPNKYOGPIPIDIVTKOAFDVTIMELICLNWTVWVEVDOSPEKINTILA 1560
 QY 1561 KINLFLVAIFTEGECIVKLAALRHYFYFTNSWNI FDEVVVILSVGTLSDDIIOKYFSPJTL 1620
 DB 1561 KINLFLVAIFTEGECIVKLAALRHYFYFTNSWNI FDEVVVILSVGTLSDDIIOKYFSPJTL 1620
 QY 1621 FRVIRIARIGRIILIRAKGIRITLLPALMNSLPALFNIGLILFVMEFYISFGMANPAY 1680
 DB 1621 FRVIRIARIGRIILIRAKGIRITLLPALMNSLPALFNIGLILFVMEFYISFGMANPAY 1680
 QY 1681 VKMEGIDDMNFQFANSMCLFOITTSAGMDGLSPLANTGPYCDPTLPNSNGSRGD 1740
 DB 1681 VKMEGIDDMNFQFANSMCLFOITTSAGMDGLSPLANTGPYCDPTLPNSNGSRGD 1740
 QY 1741 CGSPAVGLIFFTTYIIISFLIVVNMVIAIILENFVATEBESTEPESEDFFDMFYIWKXF 1800
 DB 1741 CGSPAVGLIFFTTYIIISFLIVVNMVIAIILENFVATEBESTEPESEDFFDMFYIWKXF 1800
 QY 1801 DPEAQTPIEYVLSDFADALSEPLRIAKPNQISLIMNMLPWVSGDRICHMDILFAFYKYV 1860
 DB 1801 DPEAQTPIEYVLSDFADALSEPLRIAKPNQISLIMNMLPWVSGDRICHMDILFAFYKYV 1860
 QY 1861 LGSESEMALKIOMEKEFMAANPSKISYEPIITTTLRKHEEVSANVIOQAFRRHLLQSRSL 1920
 DB 1861 LGSESEMALKIOMEKEFMAANPSKISYEPIITTTLRKHEEVSANVIOQAFRRHLLQSRSL 1920
 QY 1921 KHASELFQOAGSGISEEDAPAREGLIAYVMSSENFSPGLGPSSSSISSTSPSPSYDVT 1980
 DB 1921 KHASELFQOAGSGISEEDAPAREGLIAYVMSSENFSPGLGPSSSSISSTSPSPSYDVT 1980
 QY 1981 RATSNDNLQVRGSDYSHSEDLADPPSPDRRESIV 2015
 DB 1981 RATSNDNLQVRGSDYSHSEDLADPPSPDRRESIV 2015

RESULT 8
 ADM33995
 ID ADM33995 standard; protein: 2016 AA.
 XX
 AC ADM33995;
 XX
 DT 03-JUN-2004 (first entry)
 XX
 DE Human SCN5A variant 1 protein SEQ ID NO:2.
 XX
 KW human; cardiac sodium channel alpha subunit; SCN5A;
 KW sodium voltage-gated channel; type V; alpha; mutation study;
 KW sodium channel related disease.
 XX
 OS Homo sapiens.
 XX
 PN MO2004012668-A2.
 XX
 PD 12-FEB-2004.
 XX
 PF 01-AUG-2003; 2003WO-US024190.
 XX
 PR 02-AUG-2002; 2002US-0401018P.
 XX
 PA (WISC) WISCONSIN ALUMNI RES FOUND.
 XX
 PI Makiejski JC, Ye B, Ackerman MJ;
 XX
 DR WP1; 2004-157000/15.
 DR N-PSDB; ADM33994.
 XX
 PT New nucleic acid encoding sodium voltage-gated channel, type V, alpha
 PT polypeptide variants, useful for studying mutations, and designing or
 PT identifying new diagnostics and treatment strategies or agents for sodium
 PT channel related diseases.
 XX
 PS Claim 8; SEQ ID NO 2; 111pp; English.
 XX
 CC The present sequence represents a variant of the human cardiac sodium
 CC channel alpha subunit designated SCN5A (sodium voltage-gated channel,
 CC type V, alpha) protein. The present invention describes an isolated
 CC polynucleotide encoding an SCN5A polypeptide where the polynucleotide is
 CC selected from the group: (1) a first polynucleotide that encodes an SCN5A
 CC polypeptide selected from the group consisting of: (i) a histidine,
 CC threonine, leucine, arginine and glutamine at amino acid positions 558,
 CC 559, 618, 1027 and 1077, respectively; (ii) an arginine, threonine,
 CC 559, 618, 1027 and 1077, respectively; (iii) a histidine, threonine, leucine and
 CC arginine at amino acid positions 558, 559, 618 and 1027, respectively,
 CC with the amino acid at amino acid position 1077 deleted; or (iv) an
 CC arginine, threonine, leucine and arginine at amino acid positions 558,
 CC 559, 618 and 1027, respectively, with the amino acid at amino acid
 CC position 1077 deleted; (2) a second polynucleotide that is at least 80 %
 CC identical to the first polynucleotide over the entire length of the first
 CC polypeptide; (3) a third polynucleotide that encodes any of the SCN5A
 CC polypeptides with a conservative substitution, deletion or rearrangement
 CC at one or more non-critical amino acid position; and (4) a fourth
 CC polynucleotide that is a complement of the first, second or third
 CC polynucleotide. Also described: (1) a genetic construct comprising the
 CC polynucleotide operably linked to a non-native expression control
 CC sequence; (2) a cell comprising the polynucleotide; (3) an isolated
 CC polypeptide encoded by the polynucleotide; (4) an antibody that
 CC specifically binds to the polypeptide; (5) identifying an agent that can
 CC alter the activity of a sodium channel; (6) identifying an agent that can
 CC alter the expression of a sodium channel; (7) determining whether a
 CC biological sample or a preparation derived from the biological sample
 CC contains the polypeptide; (8) determining whether a mutation on a sodium
 CC channel is associated with a disease; and (9) determining whether a human
 CC or non-human subject is at risk for Long QT syndrome. The SCN5A
 CC polynucleotides and polypeptides are useful for studying mutations, and
 CC designing or identifying new diagnostics and treatment strategies or
 CC agents for sodium channel related diseases or conditions.
 XX

SQ	Sequence	2016	AA:																																																																																																																																																																																																																																																																																																																																																																																																																																																																																																																																																																																																																																																																																																																																																																																																																																																																																																																																																																																																																																																																																																																																																																																																																																																																																																																																																																																																																																									</
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QY	1021	PTRKETREESQPGQGTGDPBEVCPVIAVASPTDDOEDEENSLGTEESSK-0BSQ	1079						
DB	1021	PTRKETREESQPGQGTGDPBEVCPVIAVASPTDDOEDEENSLGTEESSK-0BSQ	1080						
QY	1080	PVSGGPEAPPDSTRWVSATASSSEABASQADNRQOMKPEAPAGGCTEPEDSCBSG	1139						
DB	1081	PVSGGPEAPPDSTRWVSATASSSEABASQADNRQOMKPEAPAGGCTEPEDSCBSG	1140						
QY	1140	TADMNTNELLBOIPDLQDYKDEPDCCTBCCVRRCPCCAVDTTQAPGKWMRLKTCYH	1199						
DB	1141	TADMNTNELLBOIPDLQDYKDEPDCCTBCCVRRCPCCAVDTTQAPGKWMRLKTCYH	1200						
QY	1200	IVESHWEFTFIIIFMLSSGALAFEDYLEBRKTIKVLLEYADKMFYVFLVEMLLKMYA	1259						
DB	1201	IVESHWEFTFIIIFMLSSGALAFEDYLEBRKTIKVLLEYADKMFYVFLVEMLLKMYA	1260						
QY	1260	YGPKKYFTNACWMLDFLIVDSVLSVANTLGFAEMGPISKLRTLRALRPLRALSFPFGM	1319						
DB	1261	YGPKKYFTNACWMLDFLIVDSVLSVANTLGFAEMGPISKLRTLRALRPLRALSFPFGM	1320						
QY	1320	RVVYNALVGAIPSTINVLVCLIFWLIFSINGVNLPAKRGRCINQTEGDLPLANTYVN	1379						
DB	1321	RVVYNALVGAIPSTINVLVCLIFWLIFSINGVNLPAKRGRCINQTEGDLPLANTYVN	1380						
QY	1380	KSGCESLNLGELVMTKVNFDNVGAGYLLLOVAFKGMMDIMYAAVDSRGYEOPQW	1439						
DB	1381	KSGCESLNLGELVMTKVNFDNVGAGYLLLOVAFKGMMDIMYAAVDSRGYEOPQW	1440						
QY	1440	EYNLYMYEYFIIIFGSEFTLANFIGYIDNFOQKKLGGODIIFTEBQKYYNANKK	1499						
DB	1441	EYNLYMYEYFIIIFGSEFTLANFIGYIDNFOQKKLGGODIIFTEBQKYYNANKK	1500						
QY	1500	LGSKKPOKPIPRPLANKYQGFIEDIVTKQAPDVTIMPLICLNMVTMVFETDQSPKINIL	1559						
DB	1501	LGSKKPOKPIPRPLANKYQGFIEDIVTKQAPDVTIMPLICLNMVTMVFETDQSPKINIL	1560						
QY	1560	AKINLLFVAIPTEGCIYKLAALRHVYFTNSNINIDPVVILISTYGVLSDIIOYFESPT	1619						
DB	1561	AKINLLFVAIPTEGCIYKLAALRHVYFTNSNINIDPVVILISTYGVLSDIIOYFESPT	1620						
QY	1620	LFRVIRIARIGRIILRLIRGAKGIRTLPLALMMSLPALFNIGLLFLVMFYISFGMANFA	1679						
DB	1621	LFRVIRIARIGRIILRLIRGAKGIRTLPLALMMSLPALFNIGLLFLVMFYISFGMANFA	1680						
QY	1680	VYKMEAGIDMFNFOTFANSMLCLFOITTSAGMDGLSPILANTGPYCDPTLPNSNGSRG	1739						
DB	1681	VYKMEAGIDMFNFOTFANSMLCLFOITTSAGMDGLSPILANTGPYCDPTLPNSNGSRG	1740						
QY	1740	DCGSPANGILFPTTYIIISPLIVNMATIIILENFSVATEBESTPSEDDDMYEIWEK	1799						
DB	1741	DCGSPANGILFPTTYIIISPLIVNMATIIILENFSVATEBESTPSEDDDMYEIWEK	1800						
QY	1800	FDPKATOFIEYSVLSDPADLSEPLRIAKPNOISLIMMDLPMVSGDRIHCHMDIIFAFYKR	1859						
DB	1801	FDPKATOFIEYSVLSDPADLSEPLRIAKPNOISLIMMDLPMVSGDRIHCHMDIIFAFYKR	1860						
QY	1860	VLGESGMDALKIOMEKEFMAANPSKISYEPITTTIRKRIEVSAMVIQAFRRHLLORS	1919						
DB	1861	VLGESGMDALKIOMEKEFMAANPSKISYEPITTTIRKRIEVSAMVIQAFRRHLLORS	1920						
QY	1920	LKHAASFLPROAGSGLEEDAPREBGLIAVMSNENRPLGCPRESSSISSTSPSPYDSV	1979						
DB	1921	LKHAASFLPROAGSGLEEDAPREBGLIAVMSNENRPLGCPRESSSISSTSPSPYDSV	1980						
QY	1980	TRATSDNLQVRGSDYSHSEDLADFPSPDPRDESIV 2015							
DB	1981	TRATSDNLQVRGSDYSHSEDLADFPSPDPRDESIV 2016							

RESULT 9
AEF90518
ID AEF90518 standard; protein; 2016 AA.

XX AEF90518;
AC 20-APR-2006 (first entry)
XX Human SCNS splice variant 1 SEQ ID NO 68.
DE antiarrhythmic; gene therapy; screening; diagnosis; prognosis;
XX SNP detection; genetic marker; sudden infant death syndrome; SCNS;
XX voltage-gated sodium channel.
OS Homo sapiens.
XX WO2006019984-A2.
XX 23-FEB-2006.
XX 15-JUL-2005; 2005WO-US025099.
XX 15-JUL-2004; 2004US-0588302P.
XX 14-APR-2005; 500US-05546987.
XX (UYUA) UNIV YALE.
XX Goldstein SAN, Bowers PN;
XX WPI; 2006-174087/18.
DR N-PSDB; AEF90517.
XX Testing a human individual for a marker for Sudden Infant Death Syndrome
PT comprising identifying a polymorphism in the cardiac sodium channel SCNSA
protein.
XX Claim 3; SEQ ID NO 68; 88bp; English.
XX The invention describes a method of testing a human individual for a
CC marker for Sudden Infant Death Syndrome (SIDS) comprising identifying a
CC polymorphism in a nucleic acid sample from the individual, where the
CC sample comprises both allelic copies of a nucleic acid encoding an SCNSA
CC polypeptide or copies of a protein encoded by the individual's SCNSA
CC nucleic acid. Also described are: a method of assessing a risk for SIDS
CC in a human individual; a method of identifying a human carrier of a
CC marker for SIDS; a kit, for testing a human infant or fetus for a marker,
CC assessing a risk, or for identifying a human carrier of a marker for a
CC marker for SIDS, the kit comprising: at least one reagent for identifying
CC an amino acid at a position corresponding to position 1103 in SEQ ID NO:
CC 68; and instructional material, where if the amino acid at a position
CC corresponding to position 1103 in SEQ ID NO: 2 is Y/Y, then the infant or
CC fetus has a marker for SIDS or is assessed to be at increased risk for
CC SIDS relative to an infant or fetus in which the amino acid at a position
CC corresponding to position 1103 in SEQ ID NO:68 is not Y/Y, or where if
CC the amino acid at a position corresponding to position 1103 in SEQ ID NO:
CC 68 is Y/- or Y/Y, then the human is identified as a carrier of a marker
CC for SIDS; a method of screening for SIDS in a human infant or fetus; a
CC method for diagnosing SIDS in an infant or fetus; and a method for
CC preventing SIDS in a human infant or fetus. The methods and kits are
CC useful for testing a human individual for a marker for SIDS, assessing a
CC risk for SIDS in a human individual, identifying a human carrier of a
CC marker for SIDS, and for screening and diagnosing SIDS in a human infant
CC or fetus. The method and composition are useful for preventing SIDS in a
CC human infant or fetus. This is the amino acid sequence of human SCNS
CC splice variant 1.
XX
SQ Sequence 2016 AA;
Query Match 99.9%; Score 10473.5; DB 10; Length 2016;
Best Local Similarity 99.9%; Pred No. 0;
Matches 2014; Conservative 0; Mismatches 1; Indels 1; Gaps 1;

QY SKKLDPDYGNPPOELIGEPLLEDLPFYSTOKTFVLNKGKTIFFRSATNALVYLSPPHPI 120
DB SKKLDPDYGNPPOELIGEPLLEDLPFYSTOKTFVLNKGKTIFFRSATNALVYLSPPHPI 120
QY RRAAVKILVSLFNMILMCTILTNVCVMAQHDPPPMWKYVEYFTALYTFESLVKILARG 180
DB RRAAVKILVSLFNMILMCTILTNVCVMAQHDPPPMWKYVEYFTALYTFESLVKILARG 180
QY RRAAVKILVSLFNMILMCTILTNVCVMAQHDPPPMWKYVEYFTALYTFESLVKILARG 180
DB RRAAVKILVSLFNMILMCTILTNVCVMAQHDPPPMWKYVEYFTALYTFESLVKILARG 180
QY FCLHAFPLFLADPMWMDLPSYITIMAYTTEPVDLGNVSLRFRVYRAKTISSVGLKTIIV 240
DB FCLHAFPLFLADPMWMDLPSYITIMAYTTEPVDLGNVSLRFRVYRAKTISSVGLKTIIV 240
QY FCLHAFPLFLADPMWMDLPSYITIMAYTTEPVDLGNVSLRFRVYRAKTISSVGLKTIIV 240
DB FCLHAFPLFLADPMWMDLPSYITIMAYTTEPVDLGNVSLRFRVYRAKTISSVGLKTIIV 240
QY GALIQSVKCLADVWVLTVFCLSVFALIGLQFMGNLHKCVRNFTALNGTNGSVEADGLV 300
DB GALIQSVKCLADVWVLTVFCLSVFALIGLQFMGNLHKCVRNFTALNGTNGSVEADGLV 300
QY WESIDLVLSDPENYLLKNGTSDVLLCGNSSDAGTCPEGYRCLKAGENPDHGYSFDSFAW 360
DB WESIDLVLSDPENYLLKNGTSDVLLCGNSSDAGTCPEGYRCLKAGENPDHGYSFDSFAW 360
QY APLALFPLMTQDCWERYLQOQLRSAGKIYMFPLVIFLGSFYLVNLLAVVMAAYEON 420
DB APLALFPLMTQDCWERYLQOQLRSAGKIYMFPLVIFLGSFYLVNLLAVVMAAYEON 420
QY QATIAETEERKEKRFQEMEMLKKEHEALITRGVDTVRSLSLENSPLAPVNSHERSRK 480
DB QATIAETEERKEKRFQEMEMLKKEHEALITRGVDTVRSLSLENSPLAPVNSHERSRK 480
QY RMSGTEECGEEDRLPKSDSEDPFRAMNHLSTNGLSRTSMKPRSSRSIFTPRRRDIGSE 540
DB RMSGTEECGEEDRLPKSDSEDPFRAMNHLSTNGLSRTSMKPRSSRSIFTPRRRDIGSE 540
QY RMSGTEECGEEDRLPKSDSEDPFRAMNHLSTNGLSRTSMKPRSSRSIFTPRRRDIGSE 540
DB RMSGTEECGEEDRLPKSDSEDPFRAMNHLSTNGLSRTSMKPRSSRSIFTPRRRDIGSE 540
QY ADPADDENSTAGESESRHTSLVWPPLRRTSAQQSPGTSAPGHALHGGKNSIVDQNGV 600
DB ADPADDENSTAGESESRHTSLVWPPLRRTSAQQSPGTSAPGHALHGGKNSIVDQNGV 600
QY VSLGAGDPPEATSGSHLPPVMLEHPDDTTTPEEGGQPMQLTSAQPCVDGFEEPARQ 660
DB VSLGAGDPPEATSGSHLPPVMLEHPDDTTTPEEGGQPMQLTSAQPCVDGFEEPARQ 660
QY RALGAVSLTSALELEESRRKCPCCNRLAQRVLIWECPLMWSIKQVKLVVMDPPTD 720
DB RALGAVSLTSALELEESRRKCPCCNRLAQRVLIWECPLMWSIKQVKLVVMDPPTD 720
QY LTTMCIIVLNTLPMALSHYNTSFEEMLOVGNLVFGIFPAETFKIILDPYVYQQG 780
DB LTTMCIIVLNTLPMALSHYNTSFEEMLOVGNLVFGIFPAETFKIILDPYVYQQG 780
QY WNIPIISIIIVLSLMEGLSRMSNLSVLRSPFLLRVFLKLSWPTLNTLIKIGNSVGALG 840
DB WNIPIISIIIVLSLMEGLSRMSNLSVLRSPFLLRVFLKLSWPTLNTLIKIGNSVGALG 840
QY NUTLVLAIIVIFAVVGMQLFGKNYSILRSDSGLLPRWMDFFHAFLIIFRILGEMI 900
DB NUTLVLAIIVIFAVVGMQLFGKNYSILRSDSGLLPRWMDFFHAFLIIFRILGEMI 900
QY ETWMDCHEVSGQSCLLVFLVWYTGVLVNLFLALLSFSQADNLTAPDEEMNLQ 960
DB ETWMDCHEVSGQSCLLVFLVWYTGVLVNLFLALLSFSQADNLTAPDEEMNLQ 960
QY IALARIQGLRFVRRITWDFCCGLLRQRPQKAPALAAQGLPSCIATPYSPPRETEKVP 1020
DB IALARIQGLRFVRRITWDFCCGLLRQRPQKAPALAAQGLPSCIATPYSPPRETEKVP 1020
QY PTRKETRFEGBQPGQGTGGDPEPVCPVIAVESDTDOEBDEENSIGTEBESSK-OESQ 1079
DB PTRKETRFEGBQPGQGTGGDPEPVCPVIAVESDTDOEBDEENSIGTEBESSK-OESQ 1080
QY PVRGGEPAAPPSPSRVTSQVATASSEABASASQADWRQWRAEPAPCGGTFPEBSCSGS 1139
DB PVRGGEPAAPPSPSRVTSQVATASSEABASASQADWRQWRAEPAPCGGTFPEBSCSGS 1140
QY TADMTNTNATLEBQIPDLGQDVKDEDCFTBGCVCVRCCCAVDTTQAGKQVWWRJRTCYH 1199
DB TADMTNTNATLEBQIPDLGQDVKDEDCFTBGCVCVRCCCAVDTTQAGKQVWWRJRTCYH 1199

Db 1141 TADMTNTAALBQIPDLGQDVADPEDCFEGCVRRCPCCAVDTTQAPGKVMRLKRTCH 1200
 QY 1200 IVEHSMFEFFIIIMLSSGALAIFDIYIEERKTKVLEVDKMTYFVLEMLKWA 1259
 Db 1201 IVEHSMFEFFIIIMLSSGALAIFDIYIEERKTKVLEVDKMTYFVLEMLKWA 1260
 QY 1260 YGFKKYFTNACMLDFLIVDSVLSVANTLGFAGMGPIKSLRTLRALRPLALSRFEGM 1319
 Db 1261 YGFKKYFTNACMLDFLIVDSVLSVANTLGFAGMGPIKSLRTLRALRPLALSRFEGM 1320
 QY 1320 RVVNAVALGAIPIISINNVLLVCLIFMLIFSIIMGVNLPAKRGFCINOTEGDLPLANTYVNN 1379
 Db 1321 RVVNAVALGAIPIISINNVLLVCLIFMLIFSIIMGVNLPAKRGFCINOTEGDLPLANTYVNN 1380
 QY 1380 KSQCSLNLITGELWTKVKNVDNAGAGTALLQVATFGKMDIMYAADVSRGYEQPM 1439
 Db 1381 KSQCSLNLITGELWTKVKNVDNAGAGTALLQVATFGKMDIMYAADVSRGYEQPM 1440
 QY 1440 EYNLYMYTYEVIFFIIGSFPTLNLFIGVINDPNQOKKLGQODIFMTEBOKKYTNAMK 1499
 Db 1441 EYNLYMYTYEVIFFIIGSFPTLNLFIGVINDPNQOKKLGQODIFMTEBOKKYTNAMK 1500
 QY 1500 LGSKKPQKPIPRPLNKYQGFIDIVYKQAFDVTIMFLICLNNVTMMVETDQSPKINIL 1559
 Db 1501 LGSKKPQKPIPRPLNKYQGFIDIVYKQAFDVTIMFLICLNNVTMMVETDQSPKINIL 1560
 QY 1560 AKINILFVALFTGECIVKLAALRHYYFTNSMNIPEFVVVILISIGTVLSDIIOKYFFSPT 1619
 Db 1561 AKINILFVALFTGECIVKLAALRHYYFTNSMNIPEFVVVILISIGTVLSDIIOKYFFSPT 1620
 QY 1620 LFRVRLARIGRIILIRGAKIRTLFALMMSLPLFENIGLLFLVMTYISIFMANA 1679
 Db 1621 LFRVRLARIGRIILIRGAKIRTLFALMMSLPLFENIGLLFLVMTYISIFMANA 1680
 QY 1680 YVKWAGIDDMENFOTFANSMCLFOITTSAGMDGLSPILNTGPPYCDPTLPNSNGSRG 1739
 Db 1681 YVKWAGIDDMENFOTFANSMCLFOITTSAGMDGLSPILNTGPPYCDPTLPNSNGSRG 1740
 QY 1740 DCGSAVAGILFPTTYIIISFLIVNMVYIAIILENFSVATESSTEPSDDPMFEIWEK 1799
 Db 1741 DCGSAVAGILFPTTYIIISFLIVNMVYIAIILENFSVATESSTEPSDDPMFEIWEK 1800
 QY 1800 FDBEATOFREVSUSDPAALSEPIRIKPNQISILNMDLPMVSGDRICHMDILPAFKR 1859
 Db 1801 FDBEATOFREVSUSDPAALSEPIRIKPNQISILNMDLPMVSGDRICHMDILPAFKR 1860
 QY 1860 VLGESEMDALKIOMEKFMANPSKISYEPIITTLRRKHEVSAMVIOARFRHLORS 1919
 Db 1861 VLGESEMDALKIOMEKFMANPSKISYEPIITTLRRKHEVSAMVIOARFRHLORS 1920
 QY 1920 LKHAFLFRQOAGSGLSEEDABERGLIAYVNSENSRPLGPPSSSSISSTSPSYDSV 1979
 Db 1921 LKHAFLFRQOAGSGLSEEDABERGLIAYVNSENSRPLGPPSSSSISSTSPSYDSV 1980
 QY 1980 TRATSDNLQVAGSDYSHSEDLADFPSPDRRESIV 2015
 Db 1981 TRATSDNLQVAGSDYSHSEDLADFPSPDRRESIV 2016

RESULT 10

AEA78664

ID AEA78664 standard; protein; 2015 AA.

AC AEA78664;

XX 25-AUG-2005 (first entry)

XX Human SCNSA sodium channel subunit (wild-type), SEQ ID NO:3.

XX Diagnosis; cardiovascular disease; heart arrhythmia; long QT syndrome; short QT syndrome; Brugada syndrome; progressive conduction disease; cardiac arrest; sodium channel, voltage-gated, type V, alpha; SCNSA;

KW single amino acid polymorphism; SAP.
 XX OS Homo sapiens.
 XX Key
 FH Misc-difference 104
 FT /note= "Optionally, Trp replaces wild-type Arg in a mutant SCNSA protein of the invention. Mutation is associated with Brugada syndrome"
 FT
 FT Misc-difference 179
 FT /note= "Optionally, an in-frame stop codon replaces wild-type Arg in a mutant SCNSA protein of the invention. Mutation is associated with Brugada syndrome"
 FT
 FT Misc-difference 220
 FT /note= "Optionally, Ile replaces wild-type Thr in a mutant SCNSA protein of the invention. Mutation is associated with Brugada syndrome"
 FT
 FT Misc-difference 232
 FT /note= "Optionally, Ile replaces wild-type Val in conjunction with the amino acid substitution L1307F in a mutant SCNSA protein of the invention. Mutation is associated with Brugada syndrome"
 FT
 FT Misc-difference 336
 FT /note= "Optionally, Leu replaces wild-type Pro in conjunction with the amino acid substitution I1659V in a mutant SCNSA protein of the invention. Mutation is associated with Brugada syndrome"
 FT
 FT Misc-difference 400
 FT /note= "Optionally, Ala replaces wild-type Gly in a mutant SCNSA protein of the invention. Mutation is associated with Brugada syndrome"
 FT
 FT Misc-difference 446
 FT /note= "Optionally, Lys replaces wild-type Glu in a mutant SCNSA protein of the invention. Mutation is associated with Brugada syndrome"
 FT
 FT Misc-difference 532
 FT /note= "Optionally, Cys replaces wild-type Phe in a mutant SCNSA protein of the invention. Mutation is associated with Brugada syndrome"
 FT
 FT Misc-difference 735
 FT /note= "Optionally, Val replaces wild-type Ala in a mutant SCNSA protein of the invention. Mutation is associated with Brugada syndrome"
 FT
 FT Misc-difference 851. 2015
 FT /note= "Optionally, this segment is replaced with the variant C-terminus CSSLWMACSSIAKTRTS in a mutant SCNSA protein of the invention. Mutation is associated with Brugada syndrome and results from a 2 base insertion between codons 850 and 851 (T3851 mutation)"
 FT
 FT Misc-difference 878
 FT /note= "Optionally, Cys replaces wild-type Arg in a mutant SCNSA protein of the invention. Mutation is associated with Brugada syndrome"
 FT
 FT Misc-difference 886
 FT /note= "Optionally, Pro replaces wild-type His in a mutant SCNSA protein of the invention. Mutation is associated with Brugada syndrome"
 FT
 FT Misc-difference 917
 FT /note= "Optionally, Arg replaces wild-type Leu in a mutant SCNSA protein of the invention. Mutation is associated with Brugada syndrome"
 FT
 FT Misc-difference 1008
 FT /note= "Optionally, Ser replaces wild-type Pro in a mutant SCNSA protein of the invention. Mutation is associated with progressive conduction disease"
 FT
 FT Misc-difference 1134
 FT /note= "Optionally, Ile replaces wild-type Ser in a mutant SCNSA protein of the invention. Mutation is associated with long QT syndrome"
 FT
 FT Misc-difference 1307
 FT /note= "Optionally, Phe replaces wild-type Leu in conjunction with the amino acid substitution V232I in a mutant SCNSA protein of the invention. Mutation is


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QY 841 NLTVAIAIVFIAVAVGQQLFGKNTYSELSDSDGGLPRHMMDFPHAFILIRILCGEMI 900
DB 841 NLTVAIAIVFIAVAVGQQLFGKNTYSELSDSDGGLPRHMMDFPHAFILIRILCGEMI 900
QY 901 ETMDOMEVSGSLCLVFLVAVVNLFTALLSFSADNLTADEDEMMNLQ 960
DB 901 ETMDOMEVSGSLCLVFLVAVVNLFTALLSFSADNLTADEDEMMNLQ 960
QY 961 LALANIORGLARVKTITMDPCCGLLRORPQAPALAAQOLPSCIATPYSPPEETKXP 1020
DB 961 LALANIORGLARVKTITMDPCCGLLRORPQAPALAAQOLPSCIATPYSPPEETKXP 1020
QY 1021 PTKETRFEEBEGPOGTPDEBPVCPVAVASPTDDOEDEENSLGTEEESSKQESOP 1080
DB 1021 PTKETRFEEBEGPOGTPDEBPVCPVAVASPTDDOEDEENSLGTEEESSKQESOP 1080
QY 1081 VSGGEAPPDSTRTWQSVATASSEASASQADMRQWKAEPQACGETPEPDSCEGST 1140
DB 1081 VSGGEAPPDSTRTWQSVATASSEASASQADMRQWKAEPQACGETPEPDSCEGST 1140
QY 1141 ADMNTAIELBOIPLDGDVYKDPEDCFTEGCVRCPCCAVDTTQAPGKVMRLRTCTHI 1200
DB 1141 ADMNTAIELBOIPLDGDVYKDPEDCFTEGCVRCPCCAVDTTQAPGKVMRLRTCTHI 1200
QY 1201 VEHSMETETIIFMILLSSGALAFEDIYLEERKTIKYLEVADKMTYVVLKMLKWAY 1260
DB 1201 VEHSMETETIIFMILLSSGALAFEDIYLEERKTIKYLEVADKMTYVVLKMLKWAY 1260
QY 1261 GFKKYFTNACWLDLIVDVSLVSVANTLGFENGPILKSLRTLRALPLRALSFREGMR 1320
DB 1261 GFKKYFTNACWLDLIVDVSLVSVANTLGFENGPILKSLRTLRALPLRALSFREGMR 1320
QY 1321 VVNALVGAIPISINAVLLCLIFMLIFPSIMGVNLPAKRGRCINOTEGDLPIANTYVNNK 1380
DB 1321 VVNALVGAIPISINAVLLCLIFMLIFPSIMGVNLPAKRGRCINOTEGDLPIANTYVNNK 1380
QY 1381 SOCESLNTLGEIYMKVKNPNVAGYIALLOVATPFKGMMDIMAAVDSRGYEEORPOME 1440
DB 1381 SOCESLNTLGEIYMKVKNPNVAGYIALLOVATPFKGMMDIMAAVDSRGYEEORPOME 1440
QY 1441 YNLVYIYFVFIIFIGSFFTLNLFIGVLIIDNFOQKKLGGODIMTEBOKKYANAKKL 1500
DB 1441 YNLVYIYFVFIIFIGSFFTLNLFIGVLIIDNFOQKKLGGODIMTEBOKKYANAKKL 1500
QY 1501 GSKKQKQPIPRPLNFKYQGFIDIVTKQAPDVTIMELCLINVTMMVETDDOSPEKINILIA 1560
DB 1501 GSKKQKQPIPRPLNFKYQGFIDIVTKQAPDVTIMELCLINVTMMVETDDOSPEKINILIA 1560
QY 1561 KINILFVAIFGECIVKLAALRHYYFTNSMNI FDEVVVILSVGVLSDIIOKXFFSPTL 1620
DB 1561 KINILFVAIFGECIVKLAALRHYYFTNSMNI FDEVVVILSVGVLSDIIOKXFFSPTL 1620
QY 1621 FRVIRLARIGRLIRIGAKGIRTLPLMMSLPALFNIGLLFLVMFYISIFGMANPAY 1680
DB 1621 FRVIRLARIGRLIRIGAKGIRTLPLMMSLPALFNIGLLFLVMFYISIFGMANPAY 1680
QY 1681 FRVIRLARIGRLIRIGAKGIRTLPLMMSLPALFNIGLLFLVMFYISIFGMANPAY 1680
DB 1681 FRVIRLARIGRLIRIGAKGIRTLPLMMSLPALFNIGLLFLVMFYISIFGMANPAY 1680
QY 1681 VKMEAGIDDMFNFOFTANSMLCLFOITTSAGMDGLSPILANTGPYCDPTLPNSNGSGCD 1740
DB 1681 VKMEAGIDDMFNFOFTANSMLCLFOITTSAGMDGLSPILANTGPYCDPTLPNSNGSGCD 1740
QY 1741 CGSPAVGLFTFTYIIISFLIVVMNYIAIILENFVAABEESTEPSEDDFPMFYIWEKF 1800
DB 1741 CGSPAVGLFTFTYIIISFLIVVMNYIAIILENFVAABEESTEPSEDDFPMFYIWEKF 1800
QY 1801 DPEATOFIEYSVLSDFADALSEPLRIAKPNOISLINMDLPWVSGDRICHMOILFAFTGRV 1860
DB 1801 DPEATOFIEYSVLSDFADALSEPLRIAKPNOISLINMDLPWVSGDRICHMOILFAFTGRV 1860
QY 1861 LGESGEMDALKIOMEKEMAANPSKISYEPIITTLRRKHEEVSANVIORAFRRHLLQSSL 1920
DB 1861 LGESGEMDALKIOMEKEMAANPSKISYEPIITTLRRKHEEVSANVIORAFRRHLLQSSL 1920
QY 1921 KHASLFLPQOAGSGISEEDAPBRBGLIYVMSSENSRRLGPRSSSSISSTSPPEYDSVT 1980

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DB 1921 KHASLFLPQOAGSGISEEDAPBRBGLIYVMSSENSRRLGPRSSSSISSTSPPEYDSVT 1980
QY 1981 RATSNDLQVRGSDYSHSEDLADFPSPDRRESIV 2015
DB 1981 RATSNDLQVRGSDYSHSEDLADFPSPDRRESIV 2015

RESULT 11
AAB82239
ID AAB82239 standard; protein; 2016 AA.
XX
AC AAB82239;
XX
DT 21-JUN-2001 (first entry)
XX
DE Human SCNSA protein.
XX
KW SCNSA; Long QT syndrome; LQTS; cardiovascular disease;
KW Romano-Ward syndrome; diagnosis; prognosis; therapy; drug screening.
XX
OS Homo sapiens.
XX
PN MO200124681-A2.
XX
PD 12-APR-2001.
XX
PF 09-AUG-2000; 2000WO-US021660.
XX
PR 09-AUG-1999; 99US-0147488P.
PR 17-MAR-2000; 2000US-0190057P.
XX
PA (UTAH ) UNIV UTAH RES FOUND.
XX
PI Keating MT, Splawski I;
XX
DR MPI: 2001-290564/30.
DR N-PSDB; AAF30825.
XX
PT New KVLQT1 and SCNSA genes, which contains alterations or mutations,
PT useful in diagnostic/prognostic or drug screening methods, particularly in
PT mutational analyses for screening individuals with or at risk for long QT
PT syndrome.
XX
PS Claim 31; Page 69-75; 76pp; English.
XX
CC The present sequence is that of the protein encoded by the human SCNSA
CC gene. This gene is implicated in Romano-Ward syndrome, the autosomal
CC dominant form of Long QT syndrome (LQTS). Novel mutations have been
CC identified in the gene using single strand conformation polymorphism
CC analysis. These result in the following amino acid alterations: D114N,
CC L1501V, delT1617, R1623L, E1784K and S1787N. Isolated human polypeptides
CC comprising such a mutation (see AAB82240-45) are claimed. Knowledge of
CC the mutations provides means for assessing a risk in a human subject for
CC LQTS, for diagnosing a mutation which causes LQTS, and for screening for
CC drugs useful in treating a human having a mutation in the SCNSA gene
CC
SQ
Sequence 2016 AA:
Query Match 99.5%; Score 10437.5; DB 4; Length 2016;
Best Local Similarity 99.5%; Pred. No. 0;
Matches 2006; Conservative 2; Mismatches 7; Indels 1; Gaps 1;
QY 1 MANFLPRGTSSFRFTRESIAAIEKMAEKQAGSTTLQSRGGLPEEAPRPQLDQA 60
DB 1 MANFLPRGTSSFRFTRESIAAIEKMAEKQAGSTTLQSRGGLPEEAPRPQLDQA 60
QY 61 SKKLPDIYGNPQGLISEPLEDDLPFYSTQTFIVANKKGTIFRFSATNALYVSPRPPI 120
DB 61 SKKLPDIYGNPQGLISEPLEDDLPFYSTQTFIVANKKGTIFRFSATNALYVSPRPPI 120
QY 121*GBAAVKILVSLFNNLIMCTLITNCVMAQOHDPPTTKYVEYTFATYTFESLVKILARG 180

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Db 121 RRAAVKILVHSLFNMILMCTILINCVFMAQHDPPWTKVETPTATITFESLYKILABA 180
 QY 181 FCLHAFTELRLDPWMWMLDPSVIIIMATYTBEPVDJGNVSALRTRFVRLALTKTISVIGLKTIV 240
 Db 181 FCLHAFTELRLDPWMWMLDPSVIIIMATYTBEPVDJGNVSALRTRFVRLALTKTISVIGLKTIV 240
 QY 241 GALLIOSVKKLADVWVLVTPCISVFPALLIGLOPFMGTLRHKCVRNPFALNGTNGSVBADLV 300
 Db 241 GALLIOSVKKLADVWVLVTPCISVFPALLIGLOPFMGTLRHKCVRNPFALNGTNGSVBADLV 300
 QY 301 MESIDLYSDPENYLLKNGSTDVLLCGNSSDAGTCPEGYRCLKAGENDPHGTYTSPFSAW 360
 Db 301 MESIDLYSDPENYLLKNGSTDVLLCGNSSDAGTCPEGYRCLKAGENDPHGTYTSPFSAW 360
 QY 361 APLALFRLMTODCWERLYOQTLRSAGKIYMIFFMLVIFLGSFYVNLILAVAVAMAYEBON 420
 Db 361 APLALFRLMTODCWERLYOQTLRSAGKIYMIFFMLVIFLGSFYVNLILAVAVAMAYEBON 420
 QY 421 QATTAETEEKERKPEBAMEMLKKEHEALTIRGVTVSSSLEMSPLAVNSHERSKRK 480
 Db 421 QATTAETEEKERKPEBAMEMLKKEHEALTIRGVTVSSSLEMSPLAVNSHERSKRK 480
 QY 481 RMSGTECEGDEDRLEPKSDSEDPGRAMNHLSTRGLSRTSMKPRSRSRGSIFTFRRDLGSE 540
 Db 481 RMSGTECEGDEDRLEPKSDSEDPGRAMNHLSTRGLSRTSMKPRSRSRGSIFTFRRDLGSE 540
 QY 541 ADFADDENSTAGESHSHRTSLILVWPLRRTSAQGGPSGTSAPGHALHKKONSTVDCNV 600
 Db 541 ADFADDENSTAGESHSHRTSLILVWPLRRTSAQGGPSGTSAPGHALHKKONSTVDCNV 600
 QY 601 VSLILGAGPBEATSPSSHLLRPVMLEHPDPTTTPSEBPGPQMLTSQAQCVGFEERGAQ 660
 Db 601 VSLILGAGPBEATSPSSHLLRPVMLEHPDPTTTPSEBPGPQMLTSQAQCVGFEERGAQ 660
 QY 661 RALSAYSVLTSLAELEESRHKPCPCMNRLAQRVYIWECCPLMWSIKOXVXLVWMDPPTD 720
 Db 661 RALSAYSVLTSLAELEESRHKPCPCMNRLAQRVYIWECCPLMWSIKOXVXLVWMDPPTD 720
 QY 721 LITTCIVLNTLPMALIEHNMSTSEFEEMLOVGNLVFTGIPTAEMTFKIIALDPYYFQOG 780
 Db 721 LITTCIVLNTLPMALIEHNMSTSEFEEMLOVGNLVFTGIPTAEMTFKIIALDPYYFQOG 780
 QY 781 WNIPISTIIVILSMLGLSRMSNLVLSFRLLRVFKLAKSWPTLNTLIIKIIGNSVGLG 840
 Db 781 WNIPISTIIVILSMLGLSRMSNLVLSFRLLRVFKLAKSWPTLNTLIIKIIGNSVGLG 840
 QY 841 NLTIVLAIIVFIFAVVGMLFGKNYSLELSDSGLLPRWMDPFHAFILIFRILCGEMI 900
 Db 841 NLTIVLAIIVFIFAVVGMLFGKNYSLELSDSGLLPRWMDPFHAFILIFRILCGEMI 900
 QY 901 ETMMDCEMVSQSLCLVFLVAVIGNLVNLFLALLISFSADNLTAPDEDEMANLIQ 960
 Db 901 ETMMDCEMVSQSLCLVFLVAVIGNLVNLFLALLISFSADNLTAPDEDEMANLIQ 960
 QY 961 LALATIORGLRFVXKRTTMDPCGGLRQRPQKPAALAAQOLRSCATPVSPPETEXVP 1020
 Db 961 LALATIORGLRFVXKRTTMDPCGGLRQRPQKPAALAAQOLRSCATPVSPPETEXVP 1020
 QY 1021 PTRKETRFEGEOPGOGTGPDEPEVCVPIAVAESPTDQEBDEBENSIGTEBESSK-OESQ 1079
 Db 1021 PTRKETRFEGEOPGOGTGPDEPEVCVPIAVAESPTDQEBDEBENSIGTEBESSK-OESQ 1079
 QY 1081 PVSQGPBPPDRSRMVSQVATASSEASASQAQWQKAPQAPGCGETPEDSCSEGS 1139
 Db 1081 PVSQGPBPPDRSRMVSQVATASSEASASQAQWQKAPQAPGCGETPEDSCSEGS 1139
 QY 1140 TADMNTTATLBOIPDLGQVNDPDCFTPEGVCYRRPCCAVTTTQAPGVWMLRKTQCH 1199
 Db 1140 TADMNTTATLBOIPDLGQVNDPDCFTPEGVCYRRPCCAVTTTQAPGVWMLRKTQCH 1199
 QY 1200 IYEHSMFETFIIFMILSSGALAFEDIIYEBKRTIVLLEVDKMTYIVFVLEMLKMYA 1259
 Db 1200 IYEHSMFETFIIFMILSSGALAFEDIIYEBKRTIVLLEVDKMTYIVFVLEMLKMYA 1259
 QY 1201 IYEHSMFETFIIFMILSSGALAFEDIIYEBKRTIVLLEVDKMTYIVFVLEMLKMYA 1260
 Db 1201 IYEHSMFETFIIFMILSSGALAFEDIIYEBKRTIVLLEVDKMTYIVFVLEMLKMYA 1260

QY 1260 YGPKKYFTNMACWLDLFIIVDSVLSIVANTLGAEMGPISKRLTRALRPLRALSREGM 1319
 Db 1261 YGPKKYFTNMACWLDLFIIVDSVLSIVANTLGAEMGPISKRLTRALRPLRALSREGM 1320
 QY 1320 RYVYNALVGAIPSIIMVNLVCLIFWLIIFSIMGVNLPAKFGKRCINOTEGDPLNYTIVN 1379
 Db 1321 RYVYNALVGAIPSIIMVNLVCLIFWLIIFSIMGVNLPAKFGKRCINOTEGDPLNYTIVN 1380
 QY 1380 KSQCESLNTGELVYTKVKNFNVGAGYIALLOVATPKKMNIMTAAVDSRCEBQPOW 1439
 Db 1381 KSQCESLNTGELVYTKVKNFNVGAGYIALLOVATPKKMNIMTAAVDSRCEBQPOW 1440
 QY 1440 EYVLYMYIVFVIFIFESFETLNLFGVILIDNFQOKKUGGODIFWTEBOKKYNNAMK 1499
 Db 1441 EYVLYMYIVFVIFIFESFETLNLFGVILIDNFQOKKUGGODIFWTEBOKKYNNAMK 1500
 QY 1500 LGSKKPQKPIPRPLNKYQGFIPDIVTKQAFDVTIMFLICLNVYTMVETDDQSEKINIL 1559
 Db 1501 LGSKKPQKPIPRPLNKYQGFIPDIVTKQAFDVTIMFLICLNVYTMVETDDQSEKINIL 1560
 QY 1560 AKINLFLVAIFTEGCIYKLAALRHYFTNSWNIFDFVYVILSYGTVLSDIIOKYFSPPT 1619
 Db 1561 AKINLFLVAIFTEGCIYKLAALRHYFTNSWNIFDFVYVILSYGTVLSDIIOKYFSPPT 1620
 QY 1620 LFRVIRIARIGRIILRLRGAKGIRTLIFALMWSLPALFNIGLLFLVYFISYFGMANFA 1679
 Db 1621 LFRVIRIARIGRIILRLRGAKGIRTLIFALMWSLPALFNIGLLFLVYFISYFGMANFA 1680
 QY 1680 YVKWEAGIDIMFNFQTPANSMLCLFOITTSAGMDGLSPIANTGPPYCDPTLPNSNGSRG 1739
 Db 1681 YVKWEAGIDIMFNFQTPANSMLCLFOITTSAGMDGLSPIANTGPPYCDPTLPNSNGSRG 1740
 QY 1740 DCGSPAVGILFFTTYIIISFLIVYNNKIAIILNFSVATSESTPPLSEDDPFMYETIWEK 1799
 Db 1741 DCGSPAVGILFFTTYIIISFLIVYNNKIAIILNFSVATSESTPPLSEDDPFMYETIWEK 1800
 QY 1800 FDPBATOFIEVSVDPADLSEPLRAKPNQISLIMMDLPMVSGDRHICMDILFAFTKR 1859
 Db 1801 FDPBATOFIEVSVDPADLSEPLRAKPNQISLIMMDLPMVSGDRHICMDILFAFTKR 1860
 QY 1860 VLGSSEMDALKIOBEKFMANPSKISYEBITTTLRKKEEVSAMVIOQAFRRHLLORS 1919
 Db 1861 VLGSSEMDALKIOBEKFMANPSKISYEBITTTLRKKEEVSAMVIOQAFRRHLLORS 1920
 QY 1920 LKHASFLPROQAGSLSEEDAPEREGLIAYVMSNERSPLCPBSSSISSTSPSPSYDSV 1979
 Db 1921 LKHASFLPROQAGSLSEEDAPEREGLIAYVMSNERSPLCPBSSSISSTSPSPSYDSV 1980
 QY 1980 TRATSNDLQVNGSDYSHSEDLADPPSPDRDRESIV 2015
 Db 1981 TRATSNDLQVNGSDYSHSEDLADPPSPDRDRESIV 2016

RESULT 12 4
 ADD44756
 ID ADD44756 standard; protein; 2016 AA.
 XX
 ADD44756;
 DT 29-JAN-2004 (first entry)
 XX
 DE Human Protein Q14524, SEQ ID NO 10185.
 XX
 KW Human; pain; neuronal tissue; gene therapy;
 KW spinal segmental nerve injury; chronic constriction injury; CCI;
 KW spared nerve injury; SNI; Chung.
 OS Homo sapiens.
 PN WO2003016475-A2.
 XX
 PD 27-FEB-2003.

XX 14-AUG-2002; 2002MO-US025765.
PF
XX 14-AUG-2001; 2001US-0312147P.
PR 01-NOV-2001; 2001US-0346382P.
PR 26-NOV-2001; 2001US-0333347P.
XX
XX (GHEO) GEN HOSPITAL CORP.
PA (FARB) BAYER AG.
XX
PI Woolf C, D'urso D, Befort K, Costigan M;
XX WPI; 2003-268312/26.
DR GENBANK; Q14524.
XX
PT New composition comprising two or more isolated polypeptides, useful for
XX preparing a medicament for treating pain in an animal.
PS
XX Claim 1; Page: 1017pp; English.
XX
CC The invention discloses a composition comprising two or more isolated rat
CC or human polynucleotides or a polynucleotide which represents a fragment,
CC derivative or allelic variation of the nucleic acid sequence. Also
CC claimed are a vector comprising the novel polynucleotide, a host cell
CC comprising the vector, a method for identifying a nucleotide sequence
CC which is differentially regulated in an animal subjected to pain and a
CC kit to perform the method, an array, a method for identifying an agent
CC that increases or decreases the expression of the polynucleotide sequence
CC subjected to pain, a method for identifying a compound which regulates
CC the expression of a polynucleotide sequence which is differentially
CC expressed in an animal subjected to pain, a method for identifying a
CC compound that regulates the activity of one or more of the
CC polynucleotides, a method for producing a pharmaceutical composition, a
CC method for identifying a compound or small molecule that regulates the
CC activity of one or more of the polypeptides given in the
CC specification, a method for identifying a compound useful in treating
CC pain and a pharmaceutical composition comprising the one or more
CC polypeptides or their antibodies. The polynucleotide or the compound that
CC modulates its activity is useful for preparing a medicament for treating
CC pain (e.g. spinal segmental nerve injury (Chung), chronic constriction
CC injury (CCI) and spared nerve injury (SNI)) in an animal (e.g. gene
CC therapy). The sequence presented is a human protein (shown in Table 2 of
CC the specification) which is differentially expressed during pain. Note:
CC The sequence data for this patent did not form part of the printed
CC specification, but was obtained in electronic form directly from WIPO at
CC ftp.wipo.int/pub/published_pct_sequences.
XX
SQ Sequence 2016 AA:
Query Match 99.5%; Score 10432.5; DB 7; Length 2016;
Best Local Similarity 99.5%; Pred. No. 0;
Matches 2006; Conservative 2; Mismatches 7; Indels 1; Gaps 1;
QY 1 MANPLPGTSGFRFTRESLSAIEKMAEKAQARSTLQBSRBLPEEARPOLDIOA 60
DB 1 MANPLPGTSGFRFTRESLSAIEKMAEKAQARSTLQBSRBLPEEARPOLDIOA 60
QY 61 SKKLPLDLYGNPQOELIGEPLEBDLPFYSTOKTFIYLNKQKTLFRPSATNALVYSPFPHI 120
DB 61 SKKLPLDLYGNPQOELIGEPLEBDLPFYSTOKTFIYLNKQKTLFRPSATNALVYSPFPHI 120
QY 121 RRAAVKXIIIVHSLENNLINCTIITNCVFAQAQDHPWTKYVEYTFPAITFESLVKILARG 180
DB 121 RRAAVKXIIIVHSLENNLINCTIITNCVFAQAQDHPWTKYVEYTFPAITFESLVKILARA 180
QY 181 FCLHAFETLRDPMWNLDSVITIMATTEFVUDGNVSALRTPRVVAALKTIVISLKTIV 240
DB 181 FCLHAFETLRDPMWNLDSVITIMATTEFVUDGNVSALRTPRVVAALKTIVISLKTIV 240
QY 241 GALIOSVKKLADVMVLTVFCLSVFALIGLQIFMGMLRHKCVANFTALNGTNGSVBADGLV 300
DB 241 GALIOSVKKLADVMVLTVFCLSVFALIGLQIFMGMLRHKCVANFTALNGTNGSVBADGLV 300

QY 301 WESLDLYLSDPENNYLLKNGTSDVLLCGNSSDAGTCEGGRCLKAGENPDHGYTSFDSFAM 360
DB 301 WESLDLYLSDPENNYLLKNGTSDVLLCGNSSDAGTCEGGRCLKAGENPDHGYTSFDSFAM 360
QY 361 AFLALFRMLTQDQWERYLYOQTLRSAGKIYMIFFMLVIFLGSFYLVNLLAVVAMA YBEQN 420
DB 361 AFLALFRMLTQDQWERYLYOQTLRSAGKIYMIFFMLVIFLGSFYLVNLLAVVAMA YBEQN 420
QY 421 QATTAETEKKRFPQEAEMEMLKKEHEALTITIGVTVSSLSLEMSPLAVNHSRERSKRRK 480
DB 421 QATTAETEKKRFPQEAEMEMLKKEHEALTITIGVTVSSLSLEMSPLAVNHSRERSKRRK 480
QY 481 RMSGTECGDRLPKPSDEGPRAMNLSLTRGLSRTSMKPRSRGSIFFRRRDLGSE 540
DB 481 RMSGTECGDRLPKPSDEGPRAMNLSLTRGLSRTSMKPRSRGSIFFRRRDLGSE 540
QY 541 ADPADDENSTAGSESHRTSLVWPPLRTSAQGPSPGTSAPGHALHGXKXSTVDCNGV 600
DB 541 ADPADDENSTAGSESHRTSLVWPPLRTSAQGPSPGTSAPGHALHGXKXSTVDCNGV 600
QY 601 VSLGAGDPEATSPGSHLLRPVMLEHPDPTTTPSEEPGQPMLTISOAPCVDFEEPGARQ 660
DB 601 VSLGAGDPEATSPGSHLLRPVMLEHPDPTTTPSEEPGQPMLTISOAPCVDFEEPGARQ 660
QY 661 RALSNVSVLTALBELLESRRKCPQWRLAORVLIWCCPLMMSIKQGVULVWMDPFTD 720
DB 661 RALSNVSVLTALBELLESRRKCPQWRLAORVLIWCCPLMMSIKQGVULVWMDPFTD 720
QY 721 LTTMCIYVNLTFMALHEYNMTSEFEEMLOVGNLFTGSIPTAEMTFKIALDPYVYFOG 780
DB 721 LTTMCIYVNLTFMALHEYNMTSEFEEMLOVGNLFTGSIPTAEMTFKIALDPYVYFOG 780
QY 781 WNIFPSIIIVILSLMELGLSRMSNLSVLRSPFLLRVFKLAKSWPTLNTLKIIGNSVGLG 840
DB 781 WNIFPSIIIVILSLMELGLSRMSNLSVLRSPFLLRVFKLAKSWPTLNTLKIIGNSVGLG 840
QY 841 NLTVLAIIVIFAVVGQQLFGKXYSERLSDSGLLPWHMMDPFAHLIIFRILICGMI 900
DB 841 NLTVLAIIVIFAVVGQQLFGKXYSERLSDSGLLPWHMMDPFAHLIIFRILICGMI 900
QY 901 ETMMDCHEVSGOSCLIVFLVWVIGNLVNLFALALISFSFADNLTAPEDEBEMNLQ 960
DB 901 ETMMDCHEVSGOSCLIVFLVWVIGNLVNLFALALISFSFADNLTAPEDEBEMNLQ 960
QY 961 LALARIQGLRFVKKRTIWDFFCCGLLRQRPQKPAALAAQGLPSCIATPYSPPEETEXVP 1020
DB 961 LALARIQGLRFVKKRTIWDFFCCGLLRQRPQKPAALAAQGLPSCIATPYSPPEETEXVP 1020
QY 1021 PTRKREPRFEEBOGQGTGPDPEPVCPVIAVAESPTDDQEDDENSLGTEBESSK-ORSQ 1079
DB 1021 PTRKREPRFEEBOGQGTGPDPEPVCPVIAVAESPTDDQEDDENSLGTEBESSK-ORSQ 1079
QY 1080 PVSGGPEAPPDSTRWSQVSATASSBAEASAOADWROOKAPQAGCGETPEBDCSBS 1139
DB 1080 PVSGGPEAPPDSTRWSQVSATASSBAEASAOADWROOKAPQAGCGETPEBDCSBS 1139
QY 1140 TADMNTAELLQIPLIGQDVYKDPEDCFTBSCVRRCPCCAVDTTQAPKVMRLKTCYH 1199
DB 1140 TADMNTAELLQIPLIGQDVYKDPEDCFTBSCVRRCPCCAVDTTQAPKVMRLKTCYH 1199
QY 1200 IVHSWPEFTFIIFMILSSGALAEDVYLERKKTIVLLEVAADKMFYVVFLEMLLKVA 1259
DB 1200 IVHSWPEFTFIIFMILSSGALAEDVYLERKKTIVLLEVAADKMFYVVFLEMLLKVA 1259
QY 1260 YGPKKYFTNACMIDFLIVDVSLSVANTGAPAEKGIKSLRTLRALRPLRALSREFGM 1319
DB 1260 YGPKKYFTNACMIDFLIVDVSLSVANTGAPAEKGIKSLRTLRALRPLRALSREFGM 1319
QY 1320 RVVVNAVGAIPSIIMNVLLVCLIFMLIFSINGVNLPAKFGRCINQTEGDLPLANTYVNN 1379
DB 1320 RVVVNAVGAIPSIIMNVLLVCLIFMLIFSINGVNLPAKFGRCINQTEGDLPLANTYVNN 1379
QY 1321 RVVVNAVGAIPSIIMNVLLVCLIFMLIFSINGVNLPAKFGRCINQTEGDLPLANTYVNN 1380
DB 1321 RVVVNAVGAIPSIIMNVLLVCLIFMLIFSINGVNLPAKFGRCINQTEGDLPLANTYVNN 1380

QY 1380 KSCCESLNLGTSELVTKKVNPNVAGYLLALLOVATPKGMDIMYAAVDSRGYEQPQW 1439
DB 1381 KSCCESLNLGTSELVTKKVNPNVAGYLLALLOVATPKGMDIMYAAVDSRGYEQPQW 1440
QY 1440 EYNLYMYTYPIFYIFGSEFTLNLPIGYIIDNFNOQKKLGQODIEMTEQOKKYYNMMK 1499
DB 1441 EYNLYMYTYPIFYIFGSEFTLNLPIGYIIDNFNOQKKLGQODIEMTEQOKKYYNMMK 1500
QY 1500 LGSKKPKQPIPRPLNKYQGFIDYVTKQAFVDTIMELCLNMVTMTVETDQSEPKINIL 1559
DB 1501 LGSKKPKQPIPRPLNKYQGFIDYVTKQAFVDTIMELCLNMVTMTVETDQSEPKINIL 1560
QY 1560 AKINILFPAITFGCEIVLALRHYFNNSNNIPPVVVIISYGVVSDIIOKYPFSEPT 1619
DB 1561 AKINILFPAITFGCEIVLALRHYFNNSNNIPPVVVIISYGVVSDIIOKYPFSEPT 1620
QY 1620 LFRVRLARIGRIILRIGAKGIRTLPALMMSLPALNIGLLFLVMFIYSFGMANFA 1679
DB 1621 LFRVRLARIGRIILRIGAKGIRTLPALMMSLPALNIGLLFLVMFIYSFGMANFA 1680
QY 1680 YKWBAGIDDMENFQTFANSMCLFQITTSAGWDGLSPIINTGPPYCDPTLPNSNGSRG 1739
DB 1681 YKWBAGIDDMENFQTFANSMCLFQITTSAGWDGLSPIINTGPPYCDPTLPNSNGSRG 1740
QY 1740 DCGSPAVGILPFTTYIIISPLIVNMXYAIILENFSVATBESBTEPLSEDDPMFEIMEK 1799
DB 1741 DCGSPAVGILPFTTYIIISPLIVNMXYAIILENFSVATBESBTEPLSEDDPMFEIMEK 1800
QY 1800 FDPPEATOFIEYSVLSDPADALSEPRLAKPNQISLINDLPMVSGDRHICMDILPAFTKR 1859
DB 1801 FDPPEATOFIEYSVLSDPADALSEPRLAKPNQISLINDLPMVSGDRHICMDILPAFTKR 1860
QY 1860 VLGSSEGDMDALKIOMEKFKMANPSKISYEPIITTLRKRGHEVSAMVIOQAFRRHLLORS 1919
DB 1861 VLGSSEGDMDALKIOMEKFKMANPSKISYEPIITTLRKRGHEVSAMVIOQAFRRHLLORS 1920
QY 1920 LKHAFLFRQOAGSGISEEDAPERGLAYVMSNPSPRLGPPSSSSISSTSPSPSYDSV 1979
DB 1921 LKHAFLFRQOAGSGISEEDAPERGLAYVMSNPSPRLGPPSSSSISSTSPSPSYDSV 1980
QY 1980 TRATSDNLQVRGSDYSHSEDLADFPSPSPDRRESIV 2015
DB 1981 TRATSDNLQVRGSDYSHSEDLADFPSPSPDRRESIV 2016

RESULT 13
ADE55106
ID ADE55106 strand; protein; 2016 AA.
XX ADE55106;
AC ADE55106;
XX
DT 29-JAN-2004 (first entry)
XX
DE Human Protein NP_000326, SEQ ID NO 911.
XX
KW Human; pain; neuronal tissue; gene therapy;
KW spinal segmental nerve injury; chronic constriction injury; CCI;
KW spared nerve injury; SNI; Chung.
XX
OS Homo sapiens.
XX
PN WO003016475-A2.
PD 27-FEB-2003.
XX
PF 14-AUG-2002; 2002WO-US025765.
XX
PR 14-AUG-2001; 2001US-0312147P.
PR 01-NOV-2001; 2001US-0346382P.
PR 26-NOV-2001; 2001US-033347P.
XX
PA (GEHO) GEN HOSPITAL CORP.
PA (FARB) BAYER AG.

XX
PI Woolf C, D'urso D, Befort K, Costigan M;
XX WPI; 2003-268312/26.
DR GENBANK; NP_000326.
XX
PT New composition comprising two or more isolated polypeptides, useful for
PT preparing a medicament for treating pain in an animal.
XX
PS Claim 1; Page; 1017p; English.
XX
CC The invention discloses a composition comprising two or more isolated rat
CC or human polynucleotides or a polynucleotide which represents a fragment,
CC derivative or allelic variation of the nucleic acid sequence. Also
CC comprising the vector, a method for identifying a nucleotide sequence
CC which is differentially regulated in an animal subjected to pain and a
CC kit to perform the method, an array, a method for identifying an agent
CC that increases or decreases the expression of the polynucleotide sequence
CC that is differentially expressed in neuronal tissue of a first animal
CC subjected to pain, a method for identifying a compound which regulates
CC the expression of a polynucleotide sequence which is differentially
CC expressed in an animal subjected to pain, a method for identifying a
CC compound that regulates the activity of one or more of the
CC polynucleotides, a method for producing a pharmaceutical composition, a
CC method for identifying one or more of the polypeptides given in the
CC activity in an animal of one or more of the polypeptides useful in treating
CC specification, a method for identifying a compound useful in treating
CC pain and a pharmaceutical composition comprising the one or more
CC polypeptides or their antibodies. The polynucleotide or the compound that
CC modulates its activity is useful for preparing a medicament for treating
CC pain (e.g. spinal segmental nerve injury (Chung), chronic constriction
CC injury (CCI) and spared nerve injury (SNI) in an animal (e.g. gene
CC therapy). The sequence presented is a human protein (shown in Table 2 of
CC the specification) which is differentially expressed during pain. Note:
CC The sequence data for this patent did not form part of the printed
CC specification, but was obtained in electronic form directly from WIPO at
CC ftp.wipo.int/pub/published_pcl_sequences.
SQ Sequence 2016 AA;
QY 1 MANFLLPRGSSFRFRFRESLAATEKMAEQAGSTTLDESREGLPBEEAPRQDLQA 60
DB 1 MANFLLPRGSSFRFRFRESLAATEKMAEQAGSTTLDESREGLPBEEAPRQDLQA 60
QY 61 SKKL PDL YGNPQELIGPELLEDLPFYSTOKTFIVLNKSKTIFPFSATNALYVLSPPHPI 120
DB 61 SKKL PDL YGNPQELIGPELLEDLPFYSTOKTFIVLNKSKTIFPFSATNALYVLSPPHPI 120
QY 121 RRAAVKILVHSLFNNLIMCTILNVCVMAQDHPDPMTKYVYETFTAIYTESGLYKILARG 180
DB 121 RRAAVKILVHSLFNNLIMCTILNVCVMAQDHPDPMTKYVYETFTAIYTESGLYKILARG 180
QY 121 RRAAVKILVHSLFNNLIMCTILNVCVMAQDHPDPMTKYVYETFTAIYTESGLYKILARG 180
DB 121 RRAAVKILVHSLFNNLIMCTILNVCVMAQDHPDPMTKYVYETFTAIYTESGLYKILARG 180
QY 181 FCLHAFITLADPPKMWLDPSVIMAYTTFEVDLGNVSLARTGRVRLAKTISVIGLKIYV 240
DB 181 FCLHAFITLADPPKMWLDPSVIMAYTTFEVDLGNVSLARTGRVRLAKTISVIGLKIYV 240
QY 181 FCLHAFITLADPPKMWLDPSVIMAYTTFEVDLGNVSLARTGRVRLAKTISVIGLKIYV 240
DB 181 FCLHAFITLADPPKMWLDPSVIMAYTTFEVDLGNVSLARTGRVRLAKTISVIGLKIYV 240
QY 241 GALIOSVKKGLADVWVLFYCLSVFALIGLQLEFNGNLBHKCVRNFTALNGTNGSVYADGLV 300
DB 241 GALIOSVKKGLADVWVLFYCLSVFALIGLQLEFNGNLBHKCVRNFTALNGTNGSVYADGLV 300
QY 301 WESIDLVLSDPENYLLKNGTSDVILLCGNSSDAGTCPEGYRCLKAGENDHGYTSPDPAW 360
DB 301 WESIDLVLSDPENYLLKNGTSDVILLCGNSSDAGTCPEGYRCLKAGENDHGYTSPDPAW 360
QY 361 AFALAFRLMTQDCWERLYOQTLRSAGKIYMFPMULVYFISGFYVNLILAVANAAVEEON 420
DB 361 AFALAFRLMTQDCWERLYOQTLRSAGKIYMFPMULVYFISGFYVNLILAVANAAVEEON 420

QY 421 QATIAETBEKERKFOEAMEMLKKEHEALTIRGVDIVSRSSLSMSPLAPVNSHERSKRK 480
 DB 421 QATIAETBEKERKFOEAMEMLKKEHEALTIRGVDIVSRSSLSMSPLAPVNSHERSKRK 480
 QY 481 RMSSTGEEGERLPRKSDSEDPBRAMNHLSTRGLSRTSMKPRSSRGSIFFTRRRDLGSE 540
 DB 481 RMSSTGEEGERLPRKSDSEDPBRAMNHLSTRGLSRTSMKPRSSRGSIFFTRRRDLGSE 540
 QY 541 ADEADENSTAGESHRTSLLPVMPILRRTSAQOGSPGTSAPGHALHCKKNSJTYDCNIV 600
 DB 541 ADEADENSTAGESHRTSLLPVMPILRRTSAQOGSPGTSAPGHALHCKKNSJTYDCNIV 600
 QY 601 VSLGADPEATSPSSHLLRPVMLBHPPTTTPSEBEGPQMLTQAPCVDQFEEBGAQ 660
 DB 601 VSLGADPEATSPSSHLLRPVMLBHPPTTTPSEBEGPQMLTQAPCVDQFEEBGAQ 660
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 DB 661 PALSASVITSALEBESRHKPCPCWNLQORYLIWECCLPMSIKQGVKLVMDFPTD 720
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 DB 721 LTTMCIYANTLFMALBHNMTSEFEEMLOVGNLFTGIFTAEMTFKIIALDPYVYFOQG 780
 QY 781 WNIIPDSIIIVILSLMEIGLSRMSNLSVLSFRLLRFFKLAKSWPTIANTLIIKIGNSVGLG 840
 DB 781 WNIIPDSIIIVILSLMEIGLSRMSNLSVLSFRLLRFFKLAKSWPTIANTLIIKIGNSVGLG 840
 QY 841 NITLVLAITVIFPAVAVGQOLFCKNSYSELSDSDGLPRMHMDPFAHFIIRIICGEMI 900
 DB 841 NITLVLAITVIFPAVAVGQOLFCKNSYSELSDSDGLPRMHMDPFAHFIIRIICGEMI 900
 QY 901 ETMDMCEVSGQSLCLLVFLVLMVIGNLVNLFALILSSFSANLTPADDERMNLQ 960
 DB 901 ETMDMCEVSGQSLCLLVFLVLMVIGNLVNLFALILSSFSANLTPADDERMNLQ 960
 QY 961 LALARIQGLRPFVKRTWDFCCGLRORPQKPAALAAQOLPSCIATPYSPPPETEKVP 1020
 DB 961 LALARIQGLRPFVKRTWDFCCGLRORPQKPAALAAQOLPSCIATPYSPPPETEKVP 1020
 QY 1021 PTRKETRFEEBEGPOGTFPGDEPVCPVIAVESDTDDQEDBENSIGTEBESSK-QESQ 1079
 DB 1021 PTRKETRFEEBEGPOGTFPGDEPVCPVIAVESDTDDQEDBENSIGTEBESSK-QESQ 1079
 QY 1080 PVSQGPBAPPSRRTSQQVSATSSABASASQADRWQKAPQAPGCGETPEBDCSSGS 1139
 DB 1080 PVSQGPBAPPSRRTSQQVSATSSABASASQADRWQKAPQAPGCGETPEBDCSSGS 1139
 QY 1140 TADMTNTAELLEQIPDLGQDVDPEDCFTBGCVRRCPCCAVDITQAPGKVMRLKTCYH 1199
 DB 1140 TADMTNTAELLEQIPDLGQDVDPEDCFTBGCVRRCPCCAVDITQAPGKVMRLKTCYH 1199
 QY 1200 IVESHMFETFIIFMILSSGALAFEDIYLBERTIKVILEYADKMTYVFLVLEMLLKWA 1259
 DB 1200 IVESHMFETFIIFMILSSGALAFEDIYLBERTIKVILEYADKMTYVFLVLEMLLKWA 1259
 QY 1260 YGPKKYFTNANCMDFLVVDVSVLSVANTGAPAMGPKISRTIPALRPLALSREBGM 1319
 DB 1260 YGPKKYFTNANCMDFLVVDVSVLSVANTGAPAMGPKISRTIPALRPLALSREBGM 1319
 QY 1320 RVVNAVALGAIPIINNVLLVCLIFWILFSINGVNLFAKFGRCINQTEGDLPLANTTYNN 1379
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 QY 1380 KSQCESLNLGELWYTKVKNFNDVAGYALALQVATFGKWMIDWYAAVDSRGYBEPQW 1439
 DB 1380 KSQCESLNLGELWYTKVKNFNDVAGYALALQVATFGKWMIDWYAAVDSRGYBEPQW 1439
 QY 1440 EYNLYMYTYFTIIFGSPFTLNLFTIGITINFNQOKKLGQODIEMTEBQKTYNAMK 1499
 DB 1440 EYNLYMYTYFTIIFGSPFTLNLFTIGITINFNQOKKLGQODIEMTEBQKTYNAMK 1499
 QY 1500 LGSKKPQKPIPRPLNKYGFIIDYTKQAFDVTIMFLCLANNVTMMVETDQSPKINIL 1559
 DB 1500 LGSKKPQKPIPRPLNKYGFIIDYTKQAFDVTIMFLCLANNVTMMVETDQSPKINIL 1559

DB 1501 LGSKKPQKPIPRPLNKYGFIIDYTKQAFDVTIMFLCLANNVTMMVETDQSPKINIL 1560
 QY 1560 AKINILFAIFTEGECIVLAALRHYVFTNSWNIPEPVVILISVGVVLSIDIIOKYFEEST 1619
 DB 1561 AKINILFAIFTEGECIVLAALRHYVFTNSWNIPEPVVILISVGVVLSIDIIOKYFEEST 1620
 QY 1620 LFRVIRLARIGRIILBILIGAKIRTLPLALMSLPLFNIGLLFLVMPITYSIFGMANPA 1679
 DB 1621 LFRVIRLARIGRIILBILIGAKIRTLPLALMSLPLFNIGLLFLVMPITYSIFGMANPA 1680
 QY 1680 YKMEAGIDDMENFOTFANSMCLFOITTSAGMDGLSPIANTGPPYCDPTLPSNGSRG 1739
 DB 1681 YKMEAGIDDMENFOTFANSMCLFOITTSAGMDGLSPIANTGPPYCDPTLPSNGSRG 1740
 QY 1740 DCGSPAVGILPFTTYIIISFLIVNMVYAIILENSVAETESTELSEDDPFMEIWEK 1799
 DB 1741 DCGSPAVGILPFTTYIIISFLIVNMVYAIILENSVAETESTELSEDDPFMEIWEK 1800
 QY 1800 FDPBATOFTREYSVLSDFADALSEPLRIAKPNQISLIINDLPVSGDRJHCMDILFAFTKR 1859
 DB 1801 FDPBATOFTREYSVLSDFADALSEPLRIAKPNQISLIINDLPVSGDRJHCMDILFAFTKR 1860
 QY 1860 VLGSSEMDALKIQMEKFMANPSKISYEPITTTLRKHREVSAMVIOAFRRHILQRS 1919
 DB 1861 VLGSSEMDALKIQMEKFMANPSKISYEPITTTLRKHREVSAMVIOAFRRHILQRS 1920
 QY 1920 LKHASFLEPQOAGSGLSEDDAPERGLIAYVMSNFSPPLGPSSSSISSTSPSSYSV 1979
 DB 1921 LKHASFLEPQOAGSGLSEDDAPERGLIAYVMSNFSPPLGPSSSSISSTSPSSYSV 1980
 QY 1980 TRATSNDLQVRSQVSHSEDLADPPSPDRRESTIV 2015
 DB 1981 TRATSNDLQVRSQVSHSEDLADPPSPDRRESTIV 2016

RESULT 14
 AAB82245
 ID AAB82245 standard; protein: 2016 AA.
 XX AAB82245;
 AC 21-JUN-2001 (first entry)
 DT
 XX
 DE Human SCN5A mutant S1787N.
 XX
 KW SCN5A; long QT syndrome; LQTS; cardiovascular disease;
 KW Romano-Ward syndrome; diagnosis; prognosis; therapy; drug screening;
 KW mutant; mutein.
 XX
 OS Homo sapiens.
 XX
 PN WO200124681-A2.
 XX
 PD 12-APR-2001.
 XX
 PF 09-AUG-2000; 2000WO-US021660.
 XX
 PR 09-AUG-1999; 99US-0147488P.
 PR 17-MAR-2000; 2000US-0190057P.
 XX
 PA (UTAH) UNIV UTAH RES FOUND.
 XX
 PI Keating MT, Splawski I;
 DR WPI; 2001-290564/30.
 XX
 PT New KVLQRT and SCN5A genes, which contains alterations or mutations,
 PT useful in diagnostic/prognostic or drug screening methods, particularly in
 PT mutational analyses for screening individuals with or at risk for long QT
 PT syndrome.
 XX
 PS Claim 31; Page; 76pp; English.

XX The present sequence is that of the claimed S1787N mutant of the human
CC SCNSA protein. The mutant is encoded by an SCNSA mutant gene in which a
CC G/A mutation alters codon 1787 from AGT to AAT. Mutations of the SCNSA
CC gene are implicated in Romano-Ward syndrome, the autosomal dominant form
CC of Long QT syndrome (LQTS). Mutations newly discovered in the SCNSA gene
CC lead to the following amino acid alterations in the encoded protein:
CC D114N, I1501V, delP1617, R1623L, E1784K and S1787N. Knowledge of the
CC mutations provides means for assessing a risk in a human subject for
CC LQTS, for diagnosing a mutation which causes LQTS, and for screening for
CC drugs useful in treating a human having a mutation in the SCNSA gene.
CC Note: The present sequence is not shown in the specification but is
CC derived from the KUIQT-1 sequence given in the Sequence Listing (see
CC AAB82220)
CC
CC
SQ Sequence 2016 AA;

Query Match 99.4%; Score 10429.5; DB 4; Length 2016;
Best Local Similarity 99.5%; Pred. No. 0;
Matches 2005; Conservative 3; Mismatches 7; Indels 1; Gaps 1;

QY 1 MANFLPRGTSFRFRFTRESLAIEKMAEKQARGSTTLOESREGLPREEAPRPDLQIA 60
Db 1 MANFLPRGTSFRFRFTRESLAIEKMAEKQARGSTTLOESREGLPREEAPRPDLQIA 60
QY 61 SKKLPELVGNPPQELIGLEPDLDPFYSTQKTFIVLNGKTI FRPSATNALVYLSPPHPI 120
Db 61 SKKLPELVGNPPQELIGLEPDLDPFYSTQKTFIVLNGKTI FRPSATNALVYLSPPHPI 120
QY 121 RRAVKILVHSLFNNLIMCTLTNCVMAOHDPPTVYVEYTFPAITPESLVYILARG 180
Db 121 RRAVKILVHSLFNNLIMCTLTNCVMAOHDPPTVYVEYTFPAITPESLVYILARG 180
QY 181 FCLHAFTELDPWNNLIDFSVIIIMAYTTEFVDLGNVSALRTRFRVLRALKTISVISGLKTI 240
Db 181 FCLHAFTELDPWNNLIDFSVIIIMAYTTEFVDLGNVSALRTRFRVLRALKTISVISGLKTI 240
QY 241 GALIOSVKKLADVWMLTVFCLSVFPALIGLOLFMGMLRHKCVNPFALMNGTNGSVADGLV 300
Db 241 GALIOSVKKLADVWMLTVFCLSVFPALIGLOLFMGMLRHKCVNPFALMNGTNGSVADGLV 300
QY 301 MESLDLYSDPENYLLKNGTSDVLLCGNSSDAGTPEGYRCIKAGENPDHGTSPDFPAM 360
Db 301 MESLDLYSDPENYLLKNGTSDVLLCGNSSDAGTPEGYRCIKAGENPDHGTSPDFPAM 360
QY 361 AFLALFRLMTODCWERLYOQTLRSAGKIYMFMLVIFLGSPYLVNLLAVVAMAYEON 420
Db 361 AFLALFRLMTODCWERLYOQTLRSAGKIYMFMLVIFLGSPYLVNLLAVVAMAYEON 420
QY 421 QATTIATEBEKEKRFQAMEMLKKEHEALTRGVDTVSSLSLEMSFLAPNHSHERSKRK 480
Db 421 QATTIATEBEKEKRFQAMEMLKKEHEALTRGVDTVSSLSLEMSFLAPNHSHERSKRK 480
QY 481 RMSSTEEGEGDRLLPKSDEGPRAMNHLSTFRGLSRTSMKPRSSRGSIFFTRRDDLSE 540
Db 481 RMSSTEEGEGDRLLPKSDEGPRAMNHLSTFRGLSRTSMKPRSSRGSIFFTRRDDLSE 540
QY 541 ADFADENSTAGESHRTSLVWPRLRRTSAQGPSPGTSAPGHALHGKXSTYDNCV 600
Db 541 ADFADENSTAGESHRTSLVWPRLRRTSAQGPSPGTSAPGHALHGKXSTYDNCV 600
QY 601 VSLIAGADPEATSPGSHLIRPVMLBHPDPTTTPSEBPGAPQMLTQAPCVDFEPEGARQ 660
Db 601 VSLIAGADPEATSPGSHLIRPVMLBHPDPTTTPSEBPGAPQMLTQAPCVDFEPEGARQ 660
QY 661 RALSASVYLTSLAELEESRHKCPQCMNLAQRYIIMBCCPLMMSIKQGVKLVNMPFD 720
Db 661 RALSASVYLTSLAELEESRHKCPQCMNLAQRYIIMBCCPLMMSIKQGVKLVNMPFD 720
QY 721 LTIITACIVANTLFMALIHNMTSEPEMLQVGNLFTGIFTAEMTFKIIALDPYYYPQGG 780
Db 721 LTIITACIVANTLFMALIHNMTSEPEMLQVGNLFTGIFTAEMTFKIIALDPYYYPQGG 780

QY 781 WNTFDSITVILSLMBELGRMSNLSYLRSPRLRVPFLAKSMPTLNTLKIGNSVGALG 840
Db 781 WNTFDSITVILSLMBELGRMSNLSYLRSPRLRVPFLAKSMPTLNTLKIGNSVGALG 840
QY 841 NLTIVLAIIVFPAVGMOLFGKXYSRLRSDSGLLPRTWMDPFHAEFLIIFRLIGEMI 900
Db 841 NLTIVLAIIVFPAVGMOLFGKXYSRLRSDSGLLPRTWMDPFHAEFLIIFRLIGEMI 900
QY 901 ETWMDCHVEVGQSICLVFLVNWITGNLVYLNFLALLSSFSQDNLTADDEEMNLQ 960
Db 901 ETWMDCHVEVGQSICLVFLVNWITGNLVYLNFLALLSSFSQDNLTADDEEMNLQ 960
QY 961 LALARIQGLRFVKRTTMDCCGILRQRPKPAALAAQQLPSCITATPYSPPETEKVP 1020
Db 961 LALARIQGLRFVKRTTMDCCGILRQRPKPAALAAQQLPSCITATPYSPPETEKVP 1020
QY 1021 PTRKETRPFEEGEPQGTDPDPEVCPPIVAESDTDDQDEDEBNSIGTEBESK-OESQ 1079
Db 1021 PTRKETRPFEEGEPQGTDPDPEVCPPIVAESDTDDQDEDEBNSIGTEBESK-OESQ 1080
QY 1080 PVSGGPEAPDPDSRTWSQVSATASEAASASQADWRQWKAEPQAPCCGCTPEDSCSGS 1139
Db 1081 PVSGGPEAPDPDSRTWSQVSATASEAASASQADWRQWKAEPQAPCCGCTPEDSCSGS 1140
QY 1140 TADMTNTAAILBOIPDLGQDVKDPEDCTGCVARCCCAVDTTQAPGKYWMRLRKTQYH 1199
Db 1141 TADMTNTAAILBOIPDLGQDVKDPEDCTGCVARCCCAVDTTQAPGKYWMRLRKTQYH 1200
QY 1200 IVEHSWETPFIIFMILLSSGALAEEDYLBERTIKVLEBYADMFTYVFLVEMLLKVA 1259
Db 1201 IVEHSWETPFIIFMILLSSGALAEEDYLBERTIKVLEBYADMFTYVFLVEMLLKVA 1260
QY 1260 YGFKKYFTNMCWLDPLIVDSVLSVYANTLGFEMQPISTRLRLRPRALSREGM 1319
Db 1261 YGFKKYFTNMCWLDPLIVDSVLSVYANTLGFEMQPISTRLRLRPRALSREGM 1320
QY 1320 RYVYNALVGAIPSIWNLVLCILFMLIFSLMGVLFAGKFGRCINOTEGDLPLNYTIVN 1379
Db 1321 RYVYNALVGAIPSIWNLVLCILFMLIFSLMGVLFAGKFGRCINOTEGDLPLNYTIVN 1380
QY 1380 KSQCESLNTGELYTKVNFVNVAGVYALLOVATFGKMDIMYAAVDSRGYEBQPM 1439
Db 1381 KSQCESLNTGELYTKVNFVNVAGVYALLOVATFGKMDIMYAAVDSRGYEBQPM 1440
QY 1440 EYNLYMTYPIYTFIIFSGSFYTNLFIVGIIIDNEQOKKIGGODIEMTEBOKKYNNMKX 1499
Db 1441 EYNLYMTYPIYTFIIFSGSFYTNLFIVGIIIDNEQOKKIGGODIEMTEBOKKYNNMKX 1500
QY 1500 LGSKKPQKPIPRPLNKYQGFIFDIYTRKQAFDVTIMPLICLNMVTMMVETDDQSEKINIL 1559
Db 1501 LGSKKPQKPIPRPLNKYQGFIFDIYTRKQAFDVTIMPLICLNMVTMMVETDDQSEKINIL 1560
QY 1560 AKINLLFVAIFTGECIVKLAALRHYFTNSWNIIDFVYVYIISIVGYLSDIIOXYFPSPT 1619
Db 1561 AKINLLFVAIFTGECIVKLAALRHYFTNSWNIIDFVYVYIISIVGYLSDIIOXYFPSPT 1620
QY 1620 LFRVYIRLARIRRIIRLRGAGGITLLPALMMSIPALFNGLLFLVWFYISIFGMANFA 1679
Db 1621 LFRVYIRLARIRRIIRLRGAGGITLLPALMMSIPALFNGLLFLVWFYISIFGMANFA 1680
QY 1680 YVKNBAGIDDMENFOTFANSMLCFQITTSAGMDGLSPILNTGPPYCDPTLPNSNGSRG 1739
Db 1681 YVKNBAGIDDMENFOTFANSMLCFQITTSAGMDGLSPILNTGPPYCDPTLPNSNGSRG 1740
QY 1740 DCGSPAVGILFFTYIIISFLIVNMVYAIILLENFSVATEESTEPLEDDDMYEIWEK 1799
Db 1741 DCGSPAVGILFFTYIIISFLIVNMVYAIILLENFSVATEESTEPLEDDDMYEIWEK 1800
QY 1800 FDPBEATQITRYSVSDFPADALSEPLRLAKPQOISLIMNLDPMVSGDRTHCDDILFAFVKR 1859
Db 1801 FDPBEATQITRYSVSDFPADALSEPLRLAKPQOISLIMNLDPMVSGDRTHCDDILFAFVKR 1860
QY 1860 VLGSBGENDALKIQWBEKFMANPSKISYEPITTLRKHEEVSAMVIGQAFRRHILORS 1919

DB 1861 VLGESEMDALKIQHEBEKMANPSKISYEPIITTLRRGHEVSAMVIORAFRRHLQRS 1920
QY 1920 LKHAFLPRQOAGSGISEEDAPEREGLIAYVNSEKPSRRLGPPSSSSISSTSPSPSYOSV 1979
DB 1921 LKHAFLPRQOAGSGISEEDAPEREGLIAYVNSEKPSRRLGPPSSSSISSTSPSPSYOSV 1980
QY 1980 TRATSDNLQVRGSDYSHSEDLADPPSPDRDRESIV 2015
DB 1981 TRATSDNLQVRGSDYSHSEDLADPPSPDRDRESIV 2016
RESULT 15
AAB82241
ID AAB82241 standard; protein; 2016 AA.
XX
AC AAB82241;
XX
XX 21-JUN-2001 (first entry)
DT
XX
DE Human SCNSA mutant L1501V.
XX
XX SCNSA; long QT syndrome; LQTS; cardiovascular disease;
KM Romano-Ward syndrome; diagnosis; prognosis; therapy; drug screening;
KM mutant; muteln.
OS
XX Homo sapiens.
XX
XX WO200124681-A2.
XX
XX 12-APR-2001.
XX
XX 09-AUG-2000; 2000MO-US021660.
XX
XX 09-AUG-1999; 99US-0147488P.
XX 17-MAR-2000; 2000US-0190057P.
XX
XX (UTAH) UNIV UTAH RES FOUND.
XX
XX Keating MT, Splawski I;
XX
XX WPI; 2001-290564/30.
XX
XX
XX New KVLQTI and SCNSA genes, which contains alterations or mutations.
PT useful in diagnostic/prognostic or drug screening methods, particularly in
PT mutational analyses for screening individuals with or at risk for long QT
PT syndrome.
XX
XX
XX Claim 31; Page; 76pp; English.
XX
XX The present sequence is that of the claimed L1501V mutant of the human
CC SCNSA protein. The mutant is encoded by an SCNSA mutant gene in which a
CC C/G mutation alters codon 1501 from CTG to GTG. Mutations of the SCNSA
CC gene are implicated in Romano-Ward syndrome, the autosomal dominant form
CC of long QT syndrome (LQTS). Mutations newly discovered in the SCNSA gene
CC lead to the following amino acid alterations in the encoded protein:
CC D114N, L1501V, del1617, R1623L, E1784K and S1787N. Knowledge of the
CC mutations provides means for assessing a risk in a human subject for
CC LQTS, for diagnosing a mutation which causes LQTS, and for screening for
CC drugs useful in treating a human having a mutation in the SCNSA gene.
CC Note: The present sequence is not shown in the specification but is
CC derived from the KVLQTI-1 sequence given in the Sequence Listing (see
CC AAB82240)
XX
XX
XX Sequence 2016 AA;
SQ
Query Match 99.4%; Score 10429.5; DB 4; Length 2016;
Best Local Similarity 99.5%; Pred. No. 0;
Matches 2005; Conservative 3; Mismatches 7; Indels 1; Gaps 1;

QY 1 MANFLPRGTSFRRTRESLAIIEKMAEKOARGSTTLQESREGLPEBEAPRPOLDQA 60
DB 1 MANFLPRGTSFRRTRESLAIIEKMAEKOARGSTTLQESREGLPEBEAPRPOLDQA 60

QY 61 SKGLPDLGNPQOELIGBPLEDLDPFYSTOKTFIVLNGKTIIFRESATNALVYLSPFHB 120
DB 61 SKGLPDLGNPQOELIGBPLEDLDPFYSTOKTFIVLNGKTIIFRESATNALVYLSPFHB 120
QY 121 RRAAVKIIVHSLFNMLINCTIITNCVMAQHPDPWTXYVEYTFALYTESLVKILANG 180
DB 121 RRAAVKIIVHSLFNMLINCTIITNCVMAQHPDPWTXYVEYTFALYTESLVKILANG 180
QY 181 FCLHAFYLRDPWNLDSVLIIMATTEFVLDGANSALRTFVLAALKTISVIGLKTIV 240
DB 181 FCLHAFYLRDPWNLDSVLIIMATTEFVLDGANSALRTFVLAALKTISVIGLKTIV 240
QY 241 GALLISVKKLADVWMLTVFCLSVFALIGLQFMGNLRHKCVANFTALNGTNGSVADGLV 300
DB 241 GALLISVKKLADVWMLTVFCLSVFALIGLQFMGNLRHKCVANFTALNGTNGSVADGLV 300
QY 301 WESIDLVLSDPENYLLKNGTSDVLLCGNSSDAGTCPEGYRCLKAGENDPGHYTSPDFAM 360
DB 301 WESIDLVLSDPENYLLKNGTSDVLLCGNSSDAGTCPEGYRCLKAGENDPGHYTSPDFAM 360
QY 361 AFLALFRIMTQDCWERLYQOTLRSAKTYMTFPMVLVILGSPYLVNLLAVAMAYEEN 420
DB 361 AFLALFRIMTQDCWERLYQOTLRSAKTYMTFPMVLVILGSPYLVNLLAVAMAYEEN 420
QY 421 QATTAEFEKEXRPOEAMEMLKKEHEALTIRGVDTVSRSSLEMSPLAPVNSHERSKRK 480
DB 421 QATTAEFEKEXRPOEAMEMLKKEHEALTIRGVDTVSRSSLEMSPLAPVNSHERSKRK 480
QY 481 RMSSGTECEGDRLPKSDSEDPGRAMNHLSTRGLSRTSMKRRSSRGSIFFTRRRDLGSE 540
DB 481 RMSSGTECEGDRLPKSDSEDPGRAMNHLSTRGLSRTSMKRRSSRGSIFFTRRRDLGSE 540
QY 541 ADPADDENSTAGESSHRTSLVWPPLKRTSAQCGPSGTSAFGALHGKKNSTYDCNXY 600
DB 541 ADPADDENSTAGESSHRTSLVWPPLKRTSAQCGPSGTSAFGALHGKKNSTYDCNXY 600
QY 601 VSLGAGPBPATSPESHLLRPVMLEHPDPTTPSEBPQPOLTSQAPCVDFEFGARQ 660
DB 601 VSLGAGPBPATSPESHLLRPVMLEHPDPTTPSEBPQPOLTSQAPCVDFEFGARQ 660
QY 661 RALSASVYLTALBELERHRCPCPNRLAQRYLIWECPLMWSIKOGVKLVVMDPFTD 720
DB 661 RALSASVYLTALBELERHRCPCPNRLAQRYLIWECPLMWSIKOGVKLVVMDPFTD 720
QY 721 LFTIMCIYVNLTPMALBHYNMTSEPEMLQVGNLVFTGIFPDAEMTFKIILADPYYPQOG 780
DB 721 LFTIMCIYVNLTPMALBHYNMTSEPEMLQVGNLVFTGIFPDAEMTFKIILADPYYPQOG 780
QY 781 WNIPESTIIVIIIMEIGSRMSNLSVLRSPFLLRVFKLAKSMPTLNTLIIKIIGNSVGLG 840
DB 781 WNIPESTIIVIIIMEIGSRMSNLSVLRSPFLLRVFKLAKSMPTLNTLIIKIIGNSVGLG 840
QY 841⁴MLTIVLAIIVIFAVVNGQLFGKNYSBELRDSGLIPRHHMDFFHAFLIIFRILGEMI 900
DB 841⁴MLTIVLAIIVIFAVVNGQLFGKNYSBELRDSGLIPRHHMDFFHAFLIIFRILGEMI 900
QY 901 ETMDMCEVSGQSICLLVFLVWYIGNLVINLFIALILSSPSADNLTAPDEDEMANLIQ 960
DB 901 ETMDMCEVSGQSICLLVFLVWYIGNLVINLFIALILSSPSADNLTAPDEDEMANLIQ 960
QY 961 LALARIQGLAFVVRTMDPCCGLLRORPOKPAALAAAGOLPSCICATYSPPEPTERYP 1020
DB 961 LALARIQGLAFVVRTMDPCCGLLRORPOKPAALAAAGOLPSCICATYSPPEPTERYP 1020
QY 1021 PTRKETREBEQEQGQGTGDPPEPCVPIAAESDTDDQEBDEENSLGTEBESSK-QESQ 1079
DB 1021 PTRKETREBEQEQGQGTGDPPEPCVPIAAESDTDDQEBDEENSLGTEBESSK-QESQ 1080
QY 1080 PVSQGPAPPDPSRTWSQVSATASSEAEASASQADWRQOKAPQAPGCGETPEDSCSGS 1139
DB 1081 PVSQGPAPPDPSRTWSQVSATASSEAEASASQADWRQOKAPQAPGCGETPEDSCSGS 1140

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QY 1140 TADMTNTAELLEQIPDLGQDVKDEDCETGECVRCPCCAVDTTQAPGKTMRLKTCYH 1199
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Db 1141 TADMTNTAELLEQIPDLGQDVKDEDCETGECVRCPCCAVDTTQAPGKTMRLKTCYH 1200
QY 1200 IVEHSWPEPTFIIFMILLSSGALAFEDTYLEERKTIKYULEVADKMFYVFLLEMLKMYA 1259
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Db 1201 IVEHSWPEPTFIIFMILLSSGALAFEDTYLEERKTIKYULEVADKMFYVFLLEMLKMYA 1260
QY 1260 YGFKKYFTNACWMDFLIVDSIVSLVANTLGAEMGPISKIRTLRALRPLRALSREGM 1319
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Db 1261 YGFKKYFTNACWMDFLIVDSIVSLVANTLGAEMGPISKIRTLRALRPLRALSREGM 1320
QY 1320 RVVNAVALVGAIPSIINWVLVLCIIFWLIFSIINGVNI PAKGFCRCINQTEGDLPLATTTVNN 1379
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Db 1321 RVVNAVALVGAIPSIINWVLVLCIIFWLIFSIINGVNI PAKGFCRCINQTEGDLPLATTTVNN 1380
QY 1380 KSQCESLNLGELVYTKKYNFPDNGAGYVLLAQVATKGMMDIMYAAVDSRGYEQPOW 1439
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Db 1381 KSQCESLNLGELVYTKKYNFPDNGAGYVLLAQVATKGMMDIMYAAVDSRGYEQPOW 1440
QY 1440 EYNLYMYTYFVFIIFGSGFLLNLFI GYIIDNFNOQKKKLGQODIFMTEBQKKYYNAMKK 1499
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Db 1441 EYNLYMYTYFVFIIFGSGFLLNLFI GYIIDNFNOQKKKLGQODIFMTEBQKKYYNAMKK 1500
QY 1500 LGSKKPKQKIPRPLNKKYGFIFDIYTKQAFDVTIMFLICLNMVTMMVETDQSPKINIL 1559
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Db 1501 VGSKKPKQKIPRPLNKKYGFIFDIYTKQAFDVTIMFLICLNMVTMMVETDQSPKINIL 1560
QY 1560 AKINILFVAIFMGECIVLAALRHYYPNSWNI PPFVVYVLSIVGTVSDIIOKFFSPPT 1619
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Db 1561 AKINILFVAIFMGECIVLAALRHYYPNSWNI PPFVVYVLSIVGTVSDIIOKFFSPPT 1620
QY 1620 LFRVIRLARIGRILRLIRGAKGIRTLFPALMWSLPALFNIGLLFLVNFYISIFGMANPA 1679
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Db 1621 LFRVIRLARIGRILRLIRGAKGIRTLFPALMWSLPALFNIGLLFLVNFYISIFGMANPA 1680
QY 1680 YVKWEGAGIDMENFOTFANSMLCLFOITTSAGMDGLSPILNTGPPYCDPTLPNSNGSRG 1739
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Db 1681 YVKWEGAGIDMENFOTFANSMLCLFOITTSAGMDGLSPILNTGPPYCDPTLPNSNGSRG 1740
QY 1740 DCGSPAVGLFTFTYTTIISFLVNMVYAIILLENFSVATSESTEPSLSEDDPMFYEIMBK 1799
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Db 1741 DCGSPAVGLFTFTYTTIISFLVNMVYAIILLENFSVATSESTEPSLSEDDPMFYEIMBK 1800
QY 1800 FPPKATQFIKYSVLSDFADALSEPLRIAKPNQISILINDLPWVSGDRHICMDILFAFTKR 1859
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Db 1801 FPPKATQFIKYSVLSDFADALSEPLRIAKPNQISILINDLPWVSGDRHICMDILFAFTKR 1860
QY 1860 VLGESGEMDALKIOMEKEMANPSKISYEPIITTLRRKHEVSAMVIOARFRRLHORS 1919
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Db 1861 VLGESGEMDALKIOMEKEMANPSKISYEPIITTLRRKHEVSAMVIOARFRRLHORS 1920
QY 1920 LKHAASLFRQOAGSGISEEDAPBERGLIYVMSSENRPLGPPSSSISSTSPSPSYDSV 1979
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Db 1921 LKHAASLFRQOAGSGISEEDAPBERGLIYVMSSENRPLGPPSSSISSTSPSPSYDSV 1980
QY 1980 TRATSIDLQVRSQDYSHSEDLADFPSPSPDRRESIV 2015
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Db 1981 TRATSIDLQVRSQDYSHSEDLADFPSPSPDRRESIV 2016

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KW mutant; mutein.
XX
XX Homo sapiens.
OS
XX MO200124681-A2.
PN
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XX 12-APR-2001.
PD
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XX 09-AUG-2000; 2000MO-US021660.
PF
XX
XX 09-AUG-1999; 99US-0147488P.
PR 17-MAR-2000; 2000US-0190057P.
XX
XX (UTAH ) UNIV UTAH RES FOUND.
PA
XX
XX Keating MT, Splawski I;
PI
XX WPI; 2001-290564/30.
DR
XX
XX New KYLQT1 and SCN5A gene, which contains alterations or mutations,
PT useful in diagnostic/prognostic or drug screening methods, particularly in
PT mutational analyses for screening individuals with or at risk for long QT
PT syndrome.
XX
XX
XX Claim 31; Page; 76pp; English.
XX
XX The present sequence is that of the claimed E1784K mutant of the human
CC SCN5A protein. The mutant is encoded by an SCN5A mutant gene in which a
CC G/A mutation alters codon 1784 from GAG to AAG. Mutations of the SCN5A
CC gene are implicated in Romano-Ward syndrome, the autosomal dominant form
CC of long QT syndrome (LQTS). Mutations newly discovered in the SCN5A gene
CC lead to the following amino acid alterations in the encoded protein:
CC D1114N, L1501V, delP1617, R1623L, E1784K and S1787N. Knowledge of the
CC mutations provides means for assessing a risk in a human subject for the
CC LQTS, for diagnosing a mutation which causes LQTS, and for screening for
CC drugs useful in treating a human having a mutation in the SCN5A gene.
CC Note: The present sequence is not shown in the specification but is
CC derived from the KYLQT-1 sequence given in the Sequence Listing (see
CC AAB82220)
XX
XX
XX Sequence 2016 AA;
SQ
Query Match 99.4%; Score 10428.5; DB 4; Length 2016;
Best local similarity 99.5%; Pred. No. 0;
Matches 2005; Conservative 3; Mismatches 7; Indels 1; Gaps 1;
QY 1 MANFLPRGTSFRRFTRESIALEKMAEKQARGSTTLOESRGLPEEAPRQDLQA 60
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Db 1 MANFLPRGTSFRRFTRESIALEKMAEKQARGSTTLOESRGLPEEAPRQDLQA 60
QY 61 SKKLPLDLYGNPPELIGEPLEDLPFYSTQKTFIVLNKGTIFRFSATNLVYLSPPHPI 120
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    |||
Db 61 SKKLPLDLYGNPPELIGEPLEDLPFYSTQKTFIVLNKGTIFRFSATNLVYLSPPHPI 120
QY 121 RRAAVKILVHSLFNNLIMCTIITNCVFMAQDPPEMTKYVETTAITYPESLVKILAR 180
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    |||
Db 121 RRAAVKILVHSLFNNLIMCTIITNCVFMAQDPPEMTKYVETTAITYPESLVKILAR 180
QY 181 FCLHAFTFLRDPWMLDPSVILIMAYTTEFVDLGNVSAIRTRVIRPAKLTISVIGLKTIV 240
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    |||
Db 181 FCLHAFTFLRDPWMLDPSVILIMAYTTEFVDLGNVSAIRTRVIRPAKLTISVIGLKTIV 240
QY 241 GALIOSVKKLADWVLYTFCISVPAALIGLOLFMNKILRHKCVRNFTALNGTNGSVADGLV 300
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    |||
    |||
Db 241 GALIOSVKKLADWVLYTFCISVPAALIGLOLFMNKILRHKCVRNFTALNGTNGSVADGLV 300
QY 301 WESLDLYSDPENYLLKNGTSDVLLCGNSDAGTCPEBGRCIKAGENDHGYTSDFSPAW 360
    |||
    |||
    |||
Db 301 WESLDLYSDPENYLLKNGTSDVLLCGNSDAGTCPEBGRCIKAGENDHGYTSDFSPAW 360
QY 361 AFLALFRLMTQDCWRLVQOTLRSAGKIYMIFFMLVIFLGSFYVNLILAVANAYEERON 420
    |||
    |||
    |||
Db 361 AFLALFRLMTQDCWRLVQOTLRSAGKIYMIFFMLVIFLGSFYVNLILAVANAYEERON 420

```

QY 421 QATIAETEKEKRFQFAMEMLKKEHEALTIRGVDIVSRSSLEMSPLAPVNSHERSKRK 480
 Db 421 QATIAETEKEKRFQFAMEMLKKEHEALTIRGVDIVSRSSLEMSPLAPVNSHERSKRK 480
 QY 481 RMSGTEBCEGDRLPKSDSEDPGRAMNHLSTRGLSRTSMKPRSSRGSIIFTRRRDLGSE 540
 Db 481 RMSGTEBCEGDRLPKSDSEDPGRAMNHLSTRGLSRTSMKPRSSRGSIIFTRRRDLGSE 540
 QY 541 ADPADDENSTAGESSHSRTSLVPMPLARTSAQGPSPGTSAPGALHKKNSYTDGCV 600
 Db 541 ADPADDENSTAGESSHSRTSLVPMPLARTSAQGPSPGTSAPGALHKKNSYTDGCV 600
 QY 601 VSLGAGPEATSPOSHLRPYMLEHPPTTTPSEBPGGPKMLTSOAPCVDFEPEGARQ 660
 Db 601 VSLGAGPEATSPOSHLRPYMLEHPPTTTPSEBPGGPKMLTSOAPCVDFEPEGARQ 660
 QY 661 RALSAVSVLTSALBELBESRHKCPQCNRLAQRYLIMBCCPLMMSIKQGVKLVMDDPTD 720
 Db 661 RALSAVSVLTSALBELBESRHKCPQCNRLAQRYLIMBCCPLMMSIKQGVKLVMDDPTD 720
 QY 721 LITIMCIYANTLPMALBHYNMTSEBEMLOVGNLYPTGIFTABMTFKIILADPYTFOQG 780
 Db 721 LITIMCIYANTLPMALBHYNMTSEBEMLOVGNLYPTGIFTABMTFKIILADPYTFOQG 780
 QY 781 WNIIPDSIIYIISLIMEGLSRMSNLSVLSFRLLRVPKLAKSMPITNTLIIKIIGSVGALG 840
 Db 781 WNIIPDSIIYIISLIMEGLSRMSNLSVLSFRLLRVPKLAKSMPITNTLIIKIIGSVGALG 840
 QY 841 NLTLVLAIIIVFIFAVVGMLFGKNYSBLRSDSGLLPRHMMDFPHAFIIFRILCGEVI 900
 Db 841 NLTLVLAIIIVFIFAVVGMLFGKNYSBLRSDSGLLPRHMMDFPHAFIIFRILCGEVI 900
 QY 901 ETMDMCEVSGSLCLVFLVLMVIGNLVNLFLALLISFSANLTAPEDEBEMNIQ 960
 Db 901 ETMDMCEVSGSLCLVFLVLMVIGNLVNLFLALLISFSANLTAPEDEBEMNIQ 960
 QY 961 LALARIORGLRVRKRTWDFCCGLRORPOKPAALAAQOLPSCIATPYSPPEPTEKYP 1020
 Db 961 LALARIORGLRVRKRTWDFCCGLRORPOKPAALAAQOLPSCIATPYSPPEPTEKYP 1020
 QY 1021 PTRKETREBEGPOQGTGPDPEVCPVIAVESDTDDQEBDEBENSIGTEBESSK-OESQ 1079
 Db 1021 PTRKETREBEGPOQGTGPDPEVCPVIAVESDTDDQEBDEBENSIGTEBESSK-OESQ 1079
 QY 1081 PVSQGPBPDPDSRTWSQVATASAEASASQADWRQOKAEPQAPGCGEFPEDSCSGS 1139
 Db 1081 PVSQGPBPDPDSRTWSQVATASAEASASQADWRQOKAEPQAPGCGEFPEDSCSGS 1139
 QY 1140 TADMNTAELLBOQPDLDQDVKDPEDCTEGCVRRCPCCAVDTTQAPKVMWRRLKTCYH 1199
 Db 1140 TADMNTAELLBOQPDLDQDVKDPEDCTEGCVRRCPCCAVDTTQAPKVMWRRLKTCYH 1199
 QY 1200 IVEHSMFETFLIFMLISSGALAFEDYLBERTIKVLEAYADKMTYVFLVLEMLKMYA 1259
 Db 1200 IVEHSMFETFLIFMLISSGALAFEDYLBERTIKVLEAYADKMTYVFLVLEMLKMYA 1259
 QY 1261 YGFKKFTFNAMCWDLFIVDVSLVSVANTLGFAMGPIKISIRTLARLPIRALSRFGM 1320
 Db 1261 YGFKKFTFNAMCWDLFIVDVSLVSVANTLGFAMGPIKISIRTLARLPIRALSRFGM 1320
 QY 1320 RVVNVNVAIVGATPSIINNVLLVCLIFPLISIMGVNLPACKRGCINQTEGDLPLANTYNN 1379
 Db 1320 RVVNVNVAIVGATPSIINNVLLVCLIFPLISIMGVNLPACKRGCINQTEGDLPLANTYNN 1379
 QY 1380 KSOCESLNLJGELVYTKVKNFNDVAGAYLALLQVATFGWMDIMYAADVSRGEEQPM 1439
 Db 1380 KSOCESLNLJGELVYTKVKNFNDVAGAYLALLQVATFGWMDIMYAADVSRGEEQPM 1439
 QY 1440 EYNLYMYIYFIIFGSPFTLNLFTIYIINDFNQOKKLGODIEMTEBOKKYNNAMK 1499
 Db 1440 EYNLYMYIYFIIFGSPFTLNLFTIYIINDFNQOKKLGODIEMTEBOKKYNNAMK 1499
 QY 1441 EYNLYMYIYFIIFGSPFTLNLFTIYIINDFNQOKKLGODIEMTEBOKKYNNAMK 1500
 Db 1441 EYNLYMYIYFIIFGSPFTLNLFTIYIINDFNQOKKLGODIEMTEBOKKYNNAMK 1500

QY 1500 LGSKKPQKPIPRPLNKYOGFIPIVTKQAFDVTIMFLICLANNVMTVEDDQSPKINIL 1559
 Db 1501 LGSKKPQKPIPRPLNKYOGFIPIVTKQAFDVTIMFLICLANNVMTVEDDQSPKINIL 1560
 QY 1560 AKINILFYAIFTEGICVLAALRHYYFTNSKNI EDPVVYIISIVGTVLSDIIOKTFPSPT 1619
 Db 1561 AKINILFYAIFTEGICVLAALRHYYFTNSKNI EDPVVYIISIVGTVLSDIIOKTFPSPT 1620
 QY 1620 LFRVIRLARIGRIILBIRGAGIRFTLLFALMMSLPALFNIGLLFLVMPYISIPGANA 1679
 Db 1621 LFRVIRLARIGRIILBIRGAGIRFTLLFALMMSLPALFNIGLLFLVMPYISIPGANA 1680
 QY 1680 YVKWAGIDDMENFQTFANSMCLFQITTSAGMDGLSPIINTGCPYCDPTLPNSNGSRG 1739
 Db 1681 YVKWAGIDDMENFQTFANSMCLFQITTSAGMDGLSPIINTGCPYCDPTLPNSNGSRG 1740
 QY 1740 DCGSPAVGILPFTYIIISPLVVMYIAIILENFSVATEBTEPLSEDDPMFYEIWEK 1799
 Db 1741 DCGSPAVGILPFTYIIISPLVVMYIAIILENFSVATEBTEPLSEDDPMFYEIWEK 1800
 QY 1800 FDPBATOFIEYSVLSDFADALSEPRLAKPNQISLINDLPMVSGDRJHCMDILFAFTKR 1859
 Db 1801 FDPBATOFIEYSVLSDFADALSEPRLAKPNQISLINDLPMVSGDRJHCMDILFAFTKR 1860
 QY 1860 VLGSSEMDALKIQWEEKFMANPSKISYEPIITTLARKHEVSAMVIOARFRHLORS 1919
 Db 1861 VLGSSEMDALKIQWEEKFMANPSKISYEPIITTLARKHEVSAMVIOARFRHLORS 1920
 QY 1920 LKHASFILRQAGSGLSEBAPEREGLIAYVMSNFSRPLGPSSSSISSTSPPSYDSV 1979
 Db 1921 LKHASFILRQAGSGLSEBAPEREGLIAYVMSNFSRPLGPSSSSISSTSPPSYDSV 1980
 QY 1980 TRATSNDNLOVRGSDVSHSEDLADPPSPDRRESTIV 2015
 Db 1981 TRATSNDNLOVRGSDVSHSEDLADPPSPDRRESTIV 2016

RESULT 17
 AAB82240
 ID AAB82240 standard; protein, 2016 AA.
 XX
 AC AAB82240;
 XX
 DT 21-JUN-2001 (first entry)
 XX
 DE Human SCNSA mutant D1114N.
 XX
 KW SCNSA; long QT syndrome; LQTS; cardiovascular disease;
 KW Romano-Ward syndrome; diagnosis; prognosis; therapy; drug screening;
 KW mutant; mutcin.
 XX
 OS Homo sapiens.
 XX
 PN WO200124681-A2.
 PD 12-APR-2001.
 XX
 PF 09-AUG-2000; 2000WO-US021660.
 PR 09-AUG-1999; 99US-0147488P.
 PR 17-MAR-2000; 2000US-0190057P.
 PA (UTAH) UNIV UTAH RES FOUND.
 PI Keating MT, Splawski I;
 DR WPI; 2001-290564/30.
 XX
 PT New KVLQTI and SCNSA genes, which contains alterations or mutations,
 PT useful in diagnostic/prognostic or drug screening methods, particularly in
 PT mutational analyses for screening individuals with or at risk for long QT
 PT syndrome.
 XX

QY 1860 VLGESEMDALKIOMEKEMANPSKISYEPIITTLRRKHEVSAMVIOARFRRLQRS 1919
 DB 1861 VLGESEMDALKIOMEKEMANPSKISYEPIITTLRRKHEVSAMVIOARFRRLQRS 1920
 QY 1920 LKHASFLFROQAGSGISEEDAPEREGLIAYVMSSESRPLGPPSSSSISSTSPSPSYDSV 1979
 DB 1921 LKHASFLFROQAGSGISEEDAPEREGLIAYVMSSESRPLGPPSSSSISSTSPSPSYDSV 1980
 QY 1980 TRATSDNLQVRGSDYSHSEDLADFPSPDRDRBSIV 2015
 DB 1981 TRATSDNLQVRGSDYSHSEDLADFPSPDRDRBSIV 2016

RESULT 18
 AAB82243
 ID AAB82243 standard; protein; 2016 AA.
 AC AAB82243;
 XX 21-JUN-2001 (first entry)
 DT 21-JUN-2001 (first entry)
 XX Human SCNSA mutant R1623L.
 DE
 XX SCNSA; long QT syndrome; LQTS; cardiovascular disease;
 KW Romano-Ward syndrome; diagnosis; prognosis; therapy; drug screening;
 KW mutant; mutuin.
 XX Homo sapiens.
 OS
 PN WO200124681-A2.
 PD 12-APR-2001.
 XX 09-AUG-2000; 2000WO-US021660.
 PF 09-AUG-1999; 99US-0147488P.
 PR 17-MAR-2000; 2000US-0190057P.
 XX (UTAH) UNIV UTAH RES FOUND.
 PA
 XX Keating MT, Splawski I;
 PI
 DR WPI; 2001-290564/30.
 XX
 PT New KVLQT1 and SCNSA genes, which contains alterations or mutations,
 PT useful in diagnostic/prognostic or drug screening methods, particularly in
 PT mutational analyses for screening individuals with or at risk for long QT
 PT syndrome.
 XX
 PS Claim 31; Page: 76pp; English.
 XX
 CC The present sequence is that of the claimed R1623T mutant of the human
 CC SCNSA protein. The mutant is encoded by an SCNSA mutant gene in which a
 CC G/T mutation alters codon 1623 from CGA to CTA. Mutations of the SCNSA
 CC gene are implicated in Romano-Ward syndrome, the autosomal dominant form
 CC of long QT syndrome (LQTS). Mutations newly discovered in the SCNSA gene
 CC lead to the following amino acid alterations in the encoded protein:
 CC D1114N, L1501V, delP1617, R1623L, E1784K and S1787N. Knowledge of the
 CC mutations provides means for assessing a risk in a human subject for
 CC LQTS, for diagnosing a mutation which causes LQTS, and for screening for
 CC drugs useful in treating a mutation having a mutation in the SCNSA gene.
 CC Note: The present sequence is not shown in the specification but is
 CC derived from the KVLQT-1 sequence given in the Sequence Listing (see
 CC AAB82220)
 CC
 SQ Sequence 2016 AA;

Query Match 99.4%; Score 10425.5; DB 4; Length 2016;
 Best Local Similarity 99.5%; Pred. No. 0;
 Matches 2005; Conservative 2; Mismatches 8; Indels 1; Gaps 1;

QY 1 MANFLPRTGTSFRRFTRESLIAIEKRMAEKOARGSTTLQESREGLPREEADRPOLDIOA 60
 |||

DB 1 MANFLPRTGTSFRRFTRESLIAIEKRMAEKOARGSTTLQESREGLPREEADRPOLDIOA 60
 QY 61 SKGLDLYGNPPOELIGELEDLDPFYSTQKTFIYLNKTKTFRPSATNALVLSFPHI 120
 DB 61 SKGLDLYGNPPOELIGELEDLDPFYSTQKTFIYLNKTKTFRPSATNALVLSFPHI 120
 QY 121 RRAAVKILVHSLFNNLIMCTIITNCFVMAQHPDPPTKVEYTFPAIYTFESIVKILANG 180
 DB 121 RRAAVKILVHSLFNNLIMCTIITNCFVMAQHPDPPTKVEYTFPAIYTFESIVKILANG 180
 QY 181 FCLHAFTFLRDMWNLDSVVIIMAYTFEVDIGNVSALRTFVRLAKTISVISGLKTI 240
 DB 181 FCLHAFTFLRDMWNLDSVVIIMAYTFEVDIGNVSALRTFVRLAKTISVISGLKTI 240
 QY 241 GALIOSVKKLADVNTLTPCLSVFPLIGLQFMGULRHKCVNFTALNKTNSVEADGIV 300
 DB 241 GALIOSVKKLADVNTLTPCLSVFPLIGLQFMGULRHKCVNFTALNKTNSVEADGIV 300
 QY 301 WESLDLYSDPENYLLKNGTSDVLLCGNSSDAGTCEGGRCLKAGENDPHGYSFDSFAM 360
 DB 301 WESLDLYSDPENYLLKNGTSDVLLCGNSSDAGTCEGGRCLKAGENDPHGYSFDSFAM 360
 QY 361 AFLALFRMTQDCWERYLQOTLRSAKITYMIFPMLVIFLGSFYVNLILAVVAMAYEON 420
 DB 361 AFLALFRMTQDCWERYLQOTLRSAKITYMIFPMLVIFLGSFYVNLILAVVAMAYEON 420
 QY 421 QATTIETEKEKRFQEMEMLKKEHEALTIRGVTVSSLSLEMSPLAPNSHERSKRK 480
 DB 421 QATTIETEKEKRFQEMEMLKKEHEALTIRGVTVSSLSLEMSPLAPNSHERSKRK 480
 QY 481 RWSGTEGCGDRLPKSDSDGPRAMNHLSTRGTSRTSMKRSSRGSLFTRRRDLGSE 540
 DB 481 RWSGTEGCGDRLPKSDSDGPRAMNHLSTRGTSRTSMKRSSRGSLFTRRRDLGSE 540
 QY 541 ADFADDENSTAGESESHRTSLVWPRLRTSAQGPSTGTSAGHALGKNSYDNCNV 600
 DB 541 ADFADDENSTAGESESHRTSLVWPRLRTSAQGPSTGTSAGHALGKNSYDNCNV 600
 QY 601 VSLGAGDPEATSPGSHLLRPVMLEHPDDTTTSPSEEGPGQMLTISOAPCVDFEERPARQ 660
 DB 601 VSLGAGDPEATSPGSHLLRPVMLEHPDDTTTSPSEEGPGQMLTISOAPCVDFEERPARQ 660
 QY 661 RALSASVLTALAELEESRRHKCPCMNRLAORVYIWECCPLMMSIKGVKLVWMDPFD 720
 DB 661 RALSASVLTALAELEESRRHKCPCMNRLAORVYIWECCPLMMSIKGVKLVWMDPFD 720
 QY 721 LITTCIYVNLTFMALBHYNNTSEFEEMLOVGNLVTGSIPTAEMTFKIIADPPYVPOG 780
 DB 721 LITTCIYVNLTFMALBHYNNTSEFEEMLOVGNLVTGSIPTAEMTFKIIADPPYVPOG 780
 QY 781 WNIFDSIIVILSLMELGSRMSNLSVLRSPRLRVLKAKSWPTLNTLIIKIIINSVAGLG 840
 DB 781 WNIFDSIIVILSLMELGSRMSNLSVLRSPRLRVLKAKSWPTLNTLIIKIIINSVAGLG 840
 QY 841 NUTVLATIIIVFVAVGQMLEGKNYSELKSDSGILPWHMMDPFAHLIIIFRILCGEIT 900
 DB 841 NUTVLATIIIVFVAVGQMLEGKNYSELKSDSGILPWHMMDPFAHLIIIFRILCGEIT 900
 QY 901 ETMDMDCEVSGQSCLIVFLVWVIGNLVNLFATALLSSFSADNLTAPDEBRMNNLQ 960
 DB 901 ETMDMDCEVSGQSCLIVFLVWVIGNLVNLFATALLSSFSADNLTAPDEBRMNNLQ 960
 QY 961 LALARIQGLRFVKKRTWDFCCGLIRQRPQKPAALAAQGLPSCITATYSPPPETEKYP 1020
 DB 961 LALARIQGLRFVKKRTWDFCCGLIRQRPQKPAALAAQGLPSCITATYSPPPETEKYP 1020
 QY 1021 PTRKRETEEGEOPGCGPDPBPVCVPIAAESDTDDQEDRENSLTGEESSEK-OSQ 1079
 DB 1021 PTRKRETEEGEOPGCGPDPBPVCVPIAAESDTDDQEDRENSLTGEESSEK-OSQ 1079
 QY 1080 PVSGGPEAPDPDSRTWSQVATASSEABASASQADWRQOMKAPQACGGETPEDSCSEGS 1139
 DB 1080 PVSGGPEAPDPDSRTWSQVATASSEABASASQADWRQOMKAPQACGGETPEDSCSEGS 1139
 QY 1081 PVSQMPREPPPSRTWSQVATASSEABASASQADWRQOMKAPQACGGETPEDSCSEGS 1140
 DB 1081 PVSQMPREPPPSRTWSQVATASSEABASASQADWRQOMKAPQACGGETPEDSCSEGS 1140

QY	1140	TADMTNTAELLBOJPDJGQYKDEDECTECVRCRCCCAVDTTQAQGXWMLRLKCYH	1199
Db	1141	TADMTNTAELLBOJPDJGQYKDEDECTECVRCRCCCAVDTTQAQGXWMLRLKCYH	1200
QY	1200	IYVHSWFETFIIPAILSSGALAFEDYILBERKTIKYLLLEVADRMFYVEVLEMLAKVA	1259
Db	1201	IYVHSWFETFIIPAILSSGALAFEDYILBERKTIKYLLLEVADRMFYVEVLEMLAKVA	1260
QY	1260	YGFKKYFTNACWIDLIVDSVLSVAVNTLGPVEMGPYKSLRTLRLALRPLRLSPREGM	1319
Db	1261	YGFKKYFTNACWIDLIVDSVLSVAVNTLGPVEMGPYKSLRTLRLALRPLRLSPREGM	1320
QY	1320	RYVYNALVGALPSIMNYLVCLIMWLFPSIMGVNLPGKPERCINOTEGDPLMYTLVNN	1379
Db	1321	RYVYNALVGALPSIMNYLVCLIMWLFPSIMGVNLPGKPERCINOTEGDPLMYTLVNN	1380
QY	1380	KSQCESINTLTELWYTKVKNFVDVGVGYTLALQVATEFGMMDIMYAVIDSRGYEBOPOW	1439
Db	1381	KSQCESINTLTELWYTKVKNFVDVGVGYTLALQVATEFGMMDIMYAVIDSRGYEBOPOW	1440
QY	1440	EYNLYMYIYEVYIFIIFGSFPTLNTLFQYIINFNQOKKLGODIIMTEBOKKYNNAMKK	1499
Db	1441	EYNLYMYIYEVYIFIIFGSFPTLNTLFQYIINFNQOKKLGODIIMTEBOKKYNNAMKK	1500
QY	1500	LGSKKPKQPIRPLANKOGFIFDIYTKQAFVYTIMFLICLMMYMMVETDQSEKINIL	1559
Db	1501	LGSKKPKQPIRPLANKOGFIFDIYTKQAFVYTIMFLICLMMYMMVETDQSEKINIL	1560
QY	1560	AKINILFVALFTGECIVLAALRHYYFTNSWNIFDFVNVILSIGTVLSDIIIOKYPSPPT	1619
Db	1561	AKINILFVALFTGECIVLAALRHYYFTNSWNIFDFVNVILSIGTVLSDIIIOKYPSPPT	1620
QY	1620	LFPRVIRLARIGRIIRLRIRGAKGIRTLFALMMSLPALFNIGLLFLVNFYISIFGMANFA	1679
Db	1621	LFPRVIRLARIGRIIRLRIRGAKGIRTLFALMMSLPALFNIGLLFLVNFYISIFGMANFA	1680
QY	1680	YVKNBAGIDDMFNPFOTFANSMCLFQITTSAGMDGLSPIANTGPYCDPTLPNSNGSRG	1739
Db	1681	YVKNBAGIDDMFNPFOTFANSMCLFQITTSAGMDGLSPIANTGPYCDPTLPNSNGSRG	1740
QY	1740	DCGSPAVGILFPTYIIISPLIVNMVYAIILLENFSVATESTEBLSDDPFMFEIWEK	1799
Db	1741	DCGSPAVGILFPTYIIISPLIVNMVYAIILLENFSVATESTEBLSDDPFMFEIWEK	1800
QY	1800	FDPEKTOITEYSVLSDPADLSEPLRIAKPNQOISLIMNDLPMVSGDRHCHMDIILPAFYKR	1859
Db	1801	FDPEKTOITEYSVLSDPADLSEPLRIAKPNQOISLIMNDLPMVSGDRHCHMDIILPAFYKR	1860
QY	1860	VLGSEGMDDAKIOEKEFKMAANPSKISYEPIITTLARKHEVSAWVIOQAFRHLIORS	1919
Db	1861	VLGSEGMDDAKIOEKEFKMAANPSKISYEPIITTLARKHEVSAWVIOQAFRHLIORS	1920
QY	1920	LKHASFILRQOAGSGLSEBDAEREGLIAVYVNSENFSPRLGPPSSSISSTSPSPSYSV	1979
Db	1921	LKHASFILRQOAGSGLSEBDAEREGLIAVYVNSENFSPRLGPPSSSISSTSPSPSYSV	1980
QY	1980	TRATSDNLOVAGSDYSHSEDLADPPSPDRDEESTIV	2015
Db	1981	TRATSDNLOVAGSDYSHSEDLADPPSPDRDEESTIV	2016
RESULT 19			
AAW23994	ID	AAW23994 standard; protein; 2016 AA.	
XX	AAW23994;		
XX	AC		
XX	DT	06-JUL-1998 (first entry)	
XX	DE	Human hH1 sodium channel protein.	
XX	KW	Ion channel; sodium channel; hH1; human; cardiac cell; heart; pacemaker;	

KW gene therapy.
 XX
 OS Homo sapiens.
 XX
 PN W03802040-A1.
 XX
 PD 22-JAN-1998.
 XX
 PF 04-APR-1997; 97WO-US005556.
 XX
 PR 17-JUL-1996; 96US-00682433.
 XX
 PA (MEDT) MEDTRONIC INC.
 XX
 PI Stokes KB, Norisette J;
 XX
 DR WPI; 1998-110247/10.
 DR N-PsDB; AAV09029.
 XX
 PT System for delivering genetic material to heart - comprises reservoir,
 PT catheter and optionally pacing electrode for delivering ion-channel
 PT protein, useful for, e.g. improving sensing by pacemaker.
 XX
 PS Disclosure; Page 41-47; 73pp; English.

AA This protein comprises the human h1 voltage-regulated sodium channel
CC protein that can be used in a novel system for enhancing cardiac signal
CC sensing by cardiac pacemakers through genetic treatment. A claimed system
CC for delivering genetic material (GM) comprises a reservoir containing GM
CC and a device for delivering it to myocardial cells (MC) at a specific
CC location. The GM increases the amplitude of the cardiac signal, improving
CC the signal-to-noise (S/N) ratio that is sensed by the electrode of a
CC pacemaker. Also claimed are: (1) an implantable delivery system
CC comprising a reservoir for GM which increases the expression of ion
CC channels in MC and system for delivering this through a catheter, the tip
CC of which engages MC at the chosen location, and (2) a system similar to
CC (1) comprising a pacing electrode on an inner wall of the heart, close to
CC the site where the GM is delivered. The system is used for delivery of an
CC ion-channel GM which causes depolarisation of atrial and ventricular MC
CC and improves the sensing of cardiac signals by the pacemaker and the S/N
CC ratio of atrial P-waves. The preferred GM comprises DNA (see AA09029) or
CC RNA encoding h1
CC
CC
CC
CC Sequence 2016 AA;
SQ
XX

	Score	DB 2	Length	2016
Query Match	99.4%			
Best Local Similarity	99.4%			
Best 2004, Conservative	3			
Mismatches		8		
Indels		1		
Gaps		1		

Qy	1	MANFLLPRGTSFRRPRTRESLAAIEKMAEKQARGSTLLQESREGIPEEAPRPOLDLQA	60
Db	1	MANFLLPRGTSFRRPRTRESLAAIEKMAEKQARGSTLLQESREGIPEEAPRPOLDLQA	60
Qy	61	SKKLPDLVYANPPOELIGEBLEDDPPYSTQKTFIVLANKKTI PRSATAALVYLSPFHPI	120
Db	61	SKKLPDLVYANPPOELIGEBLEDDPPYSTQKTFIVLANKKTI PRSATAALVYLSPFHPI	120
Qy	121	RRAAKVILVHSLPNNLIMCTILITNCVQMAQHP PPTWKVVEYTFPAIYTFESLVKLIARG	180
Db	121	RRAAKVILVHSLPNNLIMCTILITNCVQMAQHP PPTWKVVEYTFPAIYTFESLVKLIARA	180
Qy	181	PCJHAFTPLRDPWNMLDPSVIIIMAYTTEFVDLGNVSALRTFRVLRALKTISVISGLKTIY	240
Db	181	PCJHAFTPLRDPWNMLDPSVIIIMAYTTEFVDLGNVSALRTFRVLRALKTISVISGLKTIY	240
Qy	241	GALIOSVKKLADVMVLTVFCLSVFPALIGQLFMGNLRHKCVANFTYLANGTNSVEADGLY	300
Db	241	GALIOSVKKLADVMVLTVFCLSVFPALIGQLFMGNLRHKCVANFTYLANGTNSVEADGLV	300
Qy	301	WBSLIDLVLSDPERYLLKNGTSDVLLCGNSSDAGTCBEGYRCLKAGNPDHGYSPPDSFAM	360
Db	301	WBSLIDLVLSDPERYLLKNGTSDVLLCGNSSDAGTCBEGYRCLKAGNPDHGYSPPDSFAM	360

QY 361 AFALFRLMTOPCMBELYOOTLRSAKTYMIFFMVLVIFGSEFYVNLILAVALAMAYEON 420
 DB 361 AFALFRLMTOPCMBELYOOTLRSAKTYMIFFMVLVIFGSEFYVNLILAVALAMAYEON 420
 QY 421 QATIAETBEKEKRFQEMEMLKKHEHALTIRGVTVSRSSLEMSPLAPVNSHERSKRK 480
 DB 421 QATIAETBEKEKRFQEMEMLKKHEHALTIRGVTVSRSSLEMSPLAPVNSHERSKRK 480
 QY 481 RMSSTGEBEGEDRLPRSDSDGPRAMNHLSTRGLSRTSMKPRSSRGSIPTRRRDLSGE 540
 DB 481 RMSSTGEBEGEDRLPRSDSDGPRAMNHLSTRGLSRTSMKPRSSRGSIPTRRRDLSGE 540
 QY 541 ADFADENSTAGESHRTSLVPMPLRRTSAQOGSPGTSAPGHALHKKSTYDCNGV 600
 DB 541 ADFADENSTAGESHRTSLVPMPLRRTSAQOGSPGTSAPGHALHKKSTYDCNGV 600
 QY 601 VSLGAGDEBEATSPGSHLRLPVMLEHPDPTTTPSEBPGPOMLTSQAPCVDFEPEGARQ 660
 DB 601 VSLGAGDEBEATSPGSHLRLPVMLEHPDPTTTPSEBPGPOMLTSQAPCVDFEPEGARQ 660
 QY 661 RALSAVSUTLSALEBESRHKCPGCMNLAORYLIMECCPLMNSIKQCVKLVNMDPTD 720
 DB 661 RALSAVSUTLSALEBESRHKCPGCMNLAORYLIMECCPLMNSIKQCVKLVNMDPTD 720
 QY 721 LTTMCIVANTLPMALHEHNMTSEPEMLQVGNLVPFTGIFTAEMTFKIIALDPYYFOQG 780
 DB 721 LTTMCIVANTLPMALHEHNMTSEPEMLQVGNLVPFTGIFTAEMTFKIIALDPYYFOQG 780
 QY 781 WNIIPDSIIIVLSIMEGLSRMSNLSTVLSFLLRYFKLAKSWPTNTLTKIIGNSVGALG 840
 DB 781 WNIIPDSIIIVLSIMEGLSRMSNLSTVLSFLLRYFKLAKSWPTNTLTKIIGNSVGALG 840
 QY 841 NITLVLAIIIVFPAVVGMOLEKNTSELDSGSLPRWMMDPFAFLITRILICGEVI 900
 DB 841 NITLVLAIIIVFPAVVGMOLEKNTSELDSGSLPRWMMDPFAFLITRILICGEVI 900
 QY 901 BTMMDCEVSGOSLCLVFLVAVIGNLVNLFPLALLSSFSADNLTAPDREMNINQ 960
 DB 901 BTMMDCEVSGOSLCLVFLVAVIGNLVNLFPLALLSSFSADNLTAPDREMNINQ 960
 QY 961 LALARIORGLRFVKRTTMDFCGGLRORPQKPAALAAQOLPSCATPSPPEPEKVP 1020
 DB 961 LALARIORGLRFVKRTTMDFCGGLRORPQKPAALAAQOLPSCATPSPPEPEKVP 1020
 QY 1021 PTRKETREBEGEPQCGTIPGDEPVCVPIAVALSOTDQEBDEBNSLGTBESSK-QBSQ 1079
 DB 1021 PTRKETREBEGEPQCGTIPGDEPVCVPIAVALSOTDQEBDEBNSLGTBESSK-QBSQ 1079
 QY 1080 PVSQGPPEAPDPSRTWSQVSATASSBAASASQADWRQOKAPQAPGCGETPDEDCSBS 1139
 DB 1080 PVSQGPPEAPDPSRTWSQVSATASSBAASASQADWRQOKAPQAPGCGETPDEDCSBS 1139
 QY 1140 TADMNTNTELEBOIDPLGDVADPDCFTGECVRRCPCCAVDTTAPGKVMRLKTCYH 1199
 DB 1140 TADMNTNTELEBOIDPLGDVADPDCFTGECVRRCPCCAVDTTAPGKVMRLKTCYH 1199
 QY 1200 IVESHMPEFTIIFMILLSSGALAFEDIIYLBERTIKVILEVADKMFYVFLBMLKMYA 1259
 DB 1200 IVESHMPEFTIIFMILLSSGALAFEDIIYLBERTIKVILEVADKMFYVFLBMLKMYA 1259
 QY 1260 YGFKKFFTNAMCWLDELIVDVSVLVNNTLGFAPMGPKSLRTLRALRPLALSREFGM 1319
 DB 1260 YGFKKFFTNAMCWLDELIVDVSVLVNNTLGFAPMGPKSLRTLRALRPLALSREFGM 1319
 QY 1320 RYVVAVALVGAIPSIINNVLLVCLIFMLIFSIMGVNFAGKFGCINOTEGDPLANTYINN 1379
 DB 1320 RYVVAVALVGAIPSIINNVLLVCLIFMLIFSIMGVNFAGKFGCINOTEGDPLANTYINN 1379
 QY 1380 KSQCESLNTGELVNTKRVNFDNVAGYLLALLQVATFGKMDIMYAANDSGYEOPQW 1439
 DB 1380 KSQCESLNTGELVNTKRVNFDNVAGYLLALLQVATFGKMDIMYAANDSGYEOPQW 1439
 QY 1440 EYNLMYIYFVIFIFGSEFTLNLFTIGVILIDFNQKKKLGQDIFMTBEQKKYNNAMKX 1499

DB 1441 EYNLMYIYFVIFIFGSEFTLNLFTIGVILIDFNQKKKLGQDIFMTBEQKKYNNAMKX 1500
 QY 1500 LGSKKQKPIPRPLANKYGFIFDIYTKQAFDVTIMFLICLANNMTMMETDQSPKINIL 1559
 DB 1501 LGSKKQKPIPRPLANKYGFIFDIYTKQAFDVTIMFLICLANNMTMMETDQSPKINIL 1560
 QY 1560 AKINLFAVITGECIVLALRHYFTNSNNIPFVVVILISYGVLSIDIQKFFSPT 1619
 DB 1561 AKINLFAVITGECIVLALRHYFTNSNNIPFVVVILISYGVLSIDIQKFFSPT 1620
 QY 1620 LFRVIRLARIGRILRLINGAGIRTLPLALMMSIPALFNIGILLFLVNFYISIPGMANPA 1679
 DB 1621 LFRVIRLARIGRILRLINGAGIRTLPLALMMSIPALFNIGILLFLVNFYISIPGMANPA 1680
 QY 1680 YKWEAGIDDMFNQTFPANSMLCLFQITTSAGMDGLSPIANTGPPYCDPLPNSNGSRG 1739
 DB 1681 YKWEAGIDDMFNQTFPANSMLCLFQITTSAGMDGLSPIANTGPPYCDPLPNSNGSRG 1740
 QY 1740 DCGSPAVGILFFTYIIISPLIVNMVTAIILNFSVATBESTEPLSDDPFMEIWEK 1799
 DB 1741 DCGSPAVGILFFTYIIISPLIVNMVTAIILNFSVATBESTEPLSDDPFMEIWEK 1800
 QY 1800 FDPPEATQFIEYSVLSDFADALSEPRLAKPNQISLINDLPMVSGDRHICMDILFAFTKR 1859
 DB 1801 FDPPEATQFIEYSVLSDFADALSEPRLAKPNQISLINDLPMVSGDRHICMDILFAFTKR 1860
 QY 1860 VLGSSEMDALKIQWEEKFMANPSKISYEPIITTLRKHEBVSAMVIOAFRRHLQRS 1919
 DB 1861 VLGSSEMDALKIQWEEKFMANPSKISYEPIITTLRKHEBVSAMVIOAFRRHLQRS 1920
 QY 1920 LKHSFLLRQOAGSGLSEDEAPEREGLIAYMSENFSRPLGPSSSISSTSPSSYSV 1979
 DB 1921 LKHSFLLRQOAGSGLSEDEAPEREGLIAYMSENFSRPLGPSSSISSTSPSSYSV 1980
 QY 1980^aTRATSNDNLQVSGDYSHSEDLADPPSPDRRESIV 2015
 DB 1981 TRATSNDNLQVSGDYSHSEDLADPPSPDRRESIV 2016

RESULT 20
 AAB82242
 ID AAB82242 standard; protein, 2015 AA.
 XX AAB82242;
 AC 21-JUN-2001 (first entry)
 XX
 DT 21-JUN-2001 (first entry)
 XX
 DE Human SCNSA mutant delF1617.
 XX
 KW SCNSA; Long QT syndrome; LQTS; cardiovascular disease;
 KM Romano-Ward syndrome; diagnosis; prognosis; therapy; drug screening;
 KW mutant; mutein.
 OS Homo sapiens.
 XX
 PN W0200124681-A2.
 XX
 PD 12-APR-2001.
 XX
 PF 09-AUG-2000; 2000WO-US021660.
 XX
 PR 09-AUG-1999; 99US-0147488P.
 PR 17-MAR-2000; 2000US-0190057P.
 PA (UTAH) UNIV UTAH RES FOUND.
 XX
 PI Keating MT, Splawski I;
 XX
 DR WPI; 2001-290564/30.
 XX
 PT New KVLQTI and SCNSA genes, which contains alterations or mutations,
 useful in diagnostic/prognostic or drug screening methods, particularly in

PT mutational analyses for screening individuals with or at risk for long QT syndrome.

Claim 31; Page; 76pp; English.

CC The present sequence is that of the claimed delP617 mutant of the human
CC SCN5A protein. The mutant is encoded by an SCN5A mutant gene carrying a
CC deletion of the TTC codon for Phe-1617. Mutations of the SCN5A gene are
CC implicated in Romano-Ward syndrome, the autosomal dominant form of Long
CC QT syndrome (LQTS). Mutations newly discovered in the SCN5A gene lead to
CC the following amino acid alterations in the encoded protein: D1114N,
CC L1501V, delP617, R1623I, E1764K and S1787N. Knowledge of the mutations
CC provides means for assessing a risk in a human subject for LQTS, for
CC diagnosing a mutation which causes LQTS, and for screening for drugs
CC useful in treating a human having a mutation in the SCN5A gene. Note: The
CC present sequence is not shown in the specification but is derived from
CC the KVIQT-1 sequence given in the Sequence Listing (see AAB82220)

XX Sequence 2015 AA;

Query Match 99.3%; Score 10416; DB 4; Length 2015;

Best Local Similarity 99.5%; Pred. No. 0;

Matches 2005; Conservative 2; Mismatches 7; Indels 2; Gaps 2;

QY 1 MANFLPRTGSSFRFRTRSLAIEKMAEKQARSTTLQESREGLPREEARPQLDIOA 60
DB 1 MANFLPRTGSSFRFRTRSLAIEKMAEKQARSTTLQESREGLPREEARPQLDIOA 60
QY 61 SKKLDELGNPQOELIGEPLEDLPFYSTQKTFIYLNKSKTIFRSATNALVYLSPPFI 120
DB 61 SKKLDELGNPQOELIGEPLEDLPFYSTQKTFIYLNKSKTIFRSATNALVYLSPPFI 120
QY 121 RBAAYKILVHSLFNNLIMCTIITNCVMAQHDPPWTKXVEYTFALITYESVILARG 180
DB 121 RBAAYKILVHSLFNNLIMCTIITNCVMAQHDPPWTKXVEYTFALITYESVILARG 180
QY 181 FCLHAFPLRDPWMLDESVITMATTEFPVDIGNSALRTPFRVLRALKTISISGLKTI 240
DB 181 FCLHAFPLRDPWMLDESVITMATTEFPVDIGNSALRTPFRVLRALKTISISGLKTI 240
QY 181 FCLHAFPLRDPWMLDESVITMATTEFPVDIGNSALRTPFRVLRALKTISISGLKTI 240
DB 181 FCLHAFPLRDPWMLDESVITMATTEFPVDIGNSALRTPFRVLRALKTISISGLKTI 240
QY 241 GALLISVKKLADVMVLTVCLSVFALIGLQFMGNLRHKCVNFTALNGTNGSVEADGLV 300
DB 241 GALLISVKKLADVMVLTVCLSVFALIGLQFMGNLRHKCVNFTALNGTNGSVEADGLV 300
QY 241 GALLISVKKLADVMVLTVCLSVFALIGLQFMGNLRHKCVNFTALNGTNGSVEADGLV 300
DB 241 GALLISVKKLADVMVLTVCLSVFALIGLQFMGNLRHKCVNFTALNGTNGSVEADGLV 300
QY 301 WESLDLYSDPENYLLKNGTSDVLLCGNSSDAGTCPEGYRCLKAGBNPDHGYTSPDFAM 360
DB 301 WESLDLYSDPENYLLKNGTSDVLLCGNSSDAGTCPEGYRCLKAGBNPDHGYTSPDFAM 360
QY 301 WESLDLYSDPENYLLKNGTSDVLLCGNSSDAGTCPEGYRCLKAGBNPDHGYTSPDFAM 360
DB 301 WESLDLYSDPENYLLKNGTSDVLLCGNSSDAGTCPEGYRCLKAGBNPDHGYTSPDFAM 360
QY 361 AFLALFRLMTODCWELYYOQTLRSAGKIYMFMLVIFLGSFYLVNLLAVVAMAYEEN 420
DB 361 AFLALFRLMTODCWELYYOQTLRSAGKIYMFMLVIFLGSFYLVNLLAVVAMAYEEN 420
QY 421 QATIAETEKEKRFQAMEMLKKEHEALITRGVDYTSRSSLSEMSPLAPVNSHERSKRK 480
DB 421 QATIAETEKEKRFQAMEMLKKEHEALITRGVDYTSRSSLSEMSPLAPVNSHERSKRK 480
QY 481 RMSSGTECEGDRLPKPSDEDEGRAMNHLSTLRGLSRTSMKPRSRSRGSIFTRRDLSGE 540
DB 481 RMSSGTECEGDRLPKPSDEDEGRAMNHLSTLRGLSRTSMKPRSRSRGSIFTRRDLSGE 540
QY 481 RMSSGTECEGDRLPKPSDEDEGRAMNHLSTLRGLSRTSMKPRSRSRGSIFTRRDLSGE 540
DB 481 RMSSGTECEGDRLPKPSDEDEGRAMNHLSTLRGLSRTSMKPRSRSRGSIFTRRDLSGE 540
QY 541 ADPADDENTAGSESEHRTSLVWPMLRRTSAQGQSPGTSAGHALHKKNSYVDCNV 600
DB 541 ADPADDENTAGSESEHRTSLVWPMLRRTSAQGQSPGTSAGHALHKKNSYVDCNV 600
QY 541 ADPADDENTAGSESEHRTSLVWPMLRRTSAQGQSPGTSAGHALHKKNSYVDCNV 600
DB 541 ADPADDENTAGSESEHRTSLVWPMLRRTSAQGQSPGTSAGHALHKKNSYVDCNV 600
QY 601 VSLILGADPEATSPGSHLRPVMLRHPPTTTTSEEPGPMQLTSAQPCVDFEEBGAQ 660
DB 601 VSLILGADPEATSPGSHLRPVMLRHPPTTTTSEEPGPMQLTSAQPCVDFEEBGAQ 660
QY 661 RALSAYSVLTSALBLESRRHKPCPCMNRLAQRYLIMECCPLMWSIKQGVKLVMDPFD 720
DB 661 RALSAYSVLTSALBLESRRHKPCPCMNRLAQRYLIMECCPLMWSIKQGVKLVMDPFD 720
QY 721 LTTIMCVANTLFMALEHVMYMTSEFEBMLQVGNLVFTGIFTAEMTKIITALDPYYFQGG 780

DB 721 LTTIMCVANTLFMALEHVMYMTSEFEBMLQVGNLVFTGIFTAEMTKIITALDPYYFQGG 780
QY 781 WNIIPDSIIVILSLMEIGLSRMSNLVSRRLRLRVKLAKSWPTLNTLKIIGNSVGALG 840
DB 781 WNIIPDSIIVILSLMEIGLSRMSNLVSRRLRLRVKLAKSWPTLNTLKIIGNSVGALG 840
QY 841 NLTVLVATIFIFAVVGMOLFGKNYSERLSDSGLLPRHMDPFHAFLIIFRILGEMW 900
DB 841 NLTVLVATIFIFAVVGMOLFGKNYSERLSDSGLLPRHMDPFHAFLIIFRILGEMW 900
QY 901 ETWMDCMVEVGOSICLLVFLVWVIGNLVYVNLFLALLSSFSADNLTAPDEDEMNILQ 960
DB 901 ETWMDCMVEVGOSICLLVFLVWVIGNLVYVNLFLALLSSFSADNLTAPDEDEMNILQ 960
QY 961 LALARIQRLRFVKTITWDBCCGLLRORPQKPAALAAQGLPBCIATPYSPPEETKVP 1020
DB 961 LALARIQRLRFVKTITWDBCCGLLRORPQKPAALAAQGLPBCIATPYSPPEETKVP 1020
QY 1021 PTRKETRFEBGEOPGQTPDPPPEVCPIVAVASDTDDQEBEENSIGTEESSK-OESQ 1079
DB 1021 PTRKETRFEBGEOPGQTPDPPPEVCPIVAVASDTDDQEBEENSIGTEESSK-OESQ 1079
QY 1080 PVSGGPEAPDPSRTWSQVSATASSEASASQADWRQWAEPOAPCGGETPEDSCSEG 1139
DB 1080 PVSGGPEAPDPSRTWSQVSATASSEASASQADWRQWAEPOAPCGGETPEDSCSEG 1139
QY 1140 TADMTNTAELLBOJIPDIGNVDKDECFCTGCRCRCCQAVDTTOAGKXWMLRKTQYH 1199
DB 1140 TADMTNTAELLBOJIPDIGNVDKDECFCTGCRCRCCQAVDTTOAGKXWMLRKTQYH 1199
QY 1200 IVEHSWFETPLIFMILSSGALAFEDYIEBERKTIKYLEYADMFTYVLEMLAKVA 1259
DB 1200 IVEHSWFETPLIFMILSSGALAFEDYIEBERKTIKYLEYADMFTYVLEMLAKVA 1259
QY 1260 YGFKKFYTNAMCWLDFLIVDSLVSVANTLGFANMPKISLRTLRALRPLALSREEM 1319
DB 1260 YGFKKFYTNAMCWLDFLIVDSLVSVANTLGFANMPKISLRTLRALRPLALSREEM 1319
QY 1320 RYVNVNVALGAIPSIMNVNLVCLIFWILFSLINGVLFAGKRGRCINOTEGDPLNYTIVN 1379
DB 1320 RYVNVNVALGAIPSIMNVNLVCLIFWILFSLINGVLFAGKRGRCINOTEGDPLNYTIVN 1379
QY 1380 KSGCESLNLGELWMTYKVNFDVNGVGYALLOVAFKGMMDIMYAAVDSRGEOPW 1439
DB 1380 KSGCESLNLGELWMTYKVNFDVNGVGYALLOVAFKGMMDIMYAAVDSRGEOPW 1439
QY 1440 EYNLYMYIYFVIFIFGSPFTLNLFIGIIDFNQOKKLGQODIFMTEBOKKYNNAMK 1499
DB 1440 EYNLYMYIYFVIFIFGSPFTLNLFIGIIDFNQOKKLGQODIFMTEBOKKYNNAMK 1499
QY 1500 LGSKKPKQIPRPLNKIQGFIDIVYKQAPVYTIMPLICLNMVMTMETDQSEKINIL 1559
DB 1500 LGSKKPKQIPRPLNKIQGFIDIVYKQAPVYTIMPLICLNMVMTMETDQSEKINIL 1559
QY 1560 LGSKKPKQIPRPLNKIQGFIDIVYKQAPVYTIMPLICLNMVMTMETDQSEKINIL 1560
DB 1560 LGSKKPKQIPRPLNKIQGFIDIVYKQAPVYTIMPLICLNMVMTMETDQSEKINIL 1560
QY 1561 AKINLFPALITGCIYVLAALRHYFTNSNINIDFVYVILSIYGVLSIDLIQYK-PSPT 1619
DB 1561 AKINLFPALITGCIYVLAALRHYFTNSNINIDFVYVILSIYGVLSIDLIQYK-PSPT 1619
QY 1620 LFRVRLARIRIIRLIRGAGIRTLFLALMMSLPALFNIGLLFLVMFYISIFGMANFA 1679
DB 1620 LFRVRLARIRIIRLIRGAGIRTLFLALMMSLPALFNIGLLFLVMFYISIFGMANFA 1679
QY 1680 YVKEAGIDDMFNQTFANSMCLFOITTSAGMGLSPININTBPVCDPLPLPSNSRG 1739
DB 1680 YVKEAGIDDMFNQTFANSMCLFOITTSAGMGLSPININTBPVCDPLPLPSNSRG 1739
QY 1740 DCGSPAVGILFFTYIIISFLIVNMVYAILLENFSVATEESTEPLESDPDMYEIWEK 1799
DB 1740 DCGSPAVGILFFTYIIISFLIVNMVYAILLENFSVATEESTEPLESDPDMYEIWEK 1799
QY 1800 FDPPEATQFIEYSVLSDFADALSEPLRIAKPNOISLIMNDLPMVSGDRITHCNDILFAFTKR 1859
DB 1800 FDPPEATQFIEYSVLSDFADALSEPLRIAKPNOISLIMNDLPMVSGDRITHCNDILFAFTKR 1859

Db 1800 FDEATQPIEYSLSDPADALSEPLIAKPNQISLIMDMVSGDRHICMDILFAFTRK 1859
QY 1860 VIGSESEMDALKTOMEKEMAAAPSKISYEPIITTLARKHEEVSANVIOQAPRRHLQSS 1919
Db 1860 VIGSESEMDALKTOMEKEMAAAPSKISYEPIITTLARKHEEVSANVIOQAPRRHLQSS 1919
QY 1920 LKHASFLFRQOAGSGISEBDAPEREGLIAYVMSSENSRPLGPPSSSSISSTSPPSYDSV 1979
Db 1920 LKHASFLFRQOAGSGISEBDAPEREGLIAYVMSSENSRPLGPPSSSSISSTSPPSYDSV 1979
QY 1980 TRATSNDLVGRSGDYSHSEDLADPPSPDRRESIV 2015
Db 1980 TRATSNDLVGRSGDYSHSEDLADPPSPDRRESIV 2015

RESULT 21
AAR67913
ID AAR67913 standard; protein; 2019 AA.
XX AAR67913;
AC 25-MAR-2003 (revised)
DT 05-AUG-1995 (first entry)
XX Cardiac sodium channel protein.
XX Sodium channel protein; therapeutic; diagnostic; prognostic;
XX anclarythmic; cardiant; cardioglycoside.
XX Rattus rattus.
XX US5380836-A.
XX 10-JAN-1995.
XX 30-SEP-1991; 91US-00768107.
XX 13-FEB-1989; 89US-00331330.
XX (ARCH-) ARCH DEV CORP.
XX Rogart RB;
XX MPI: 1995-060381/08.
XX P-PsDB; AA081328.
XX Purified DNA's encoding rat and human cardiac sodium channel protein -
XX useful for recombinant expression to produce sodium channel proteins.
XX Disclosure; Fig 2; 39pp; English.
XX The rat cardiac channel protein has various therapeutic, diagnostic and
XX prognostic uses. It may also be used to develop more effective
XX anclarythmic, cardiant and cardioglycoside drugs. In Figure 2, the
XX sequence is compared to the deduced amino acid sequence of rat brain II
XX cDNA. (Updated on 25-MAR-2003 to correct PF field.)
SQ Sequence 2019 AA;

Query Match 93.1%; Score 9767; DB 2; Length 2019;
Best Local Similarity 93.3%; Pred. No. 0;
Matches 1884; Conservative 47; Mismatches 84; Indels 4; Gaps 3;

QY 1 MANFLPRTGSSFRRTRESLAIEKMAEKQAR-GSTTLQSSREGLPPEBAAPRQDLQ 59
Db 1 MANFLPRTGSSFRRTRESLAIEKMAEKQAR-GSTTLQSSREGLPPEBAAPRQDLQ 60
QY 60 ASKKLPDLXGNPPELIGLEPDLDPFYSTOKTIFVANKGKTIIPRSATNAYVSPFRP 119
Db 61 ASKKLPDLXGNPPELIGLEPDLDPFYSTOKTIFVANKGKTIIPRSATNAYVSPFRP 120
QY 120 IRRRAVKILVHSLFMNLIWCTILTNCFVMAQHPPEWTKYVEYTFATYTFESLVKILAR 179

Db 121 VRRAAVKILVHSLFMNLIWCTILTNCFVMAQHPPEWTKYVEYTFATYTFESLVKILAR 180
QY 180 GCLIAHFTLRDPPMMMLDSSVILIMATTEFVULGNVSAIRTRVIALATISVIGLKI 239
Db 181 GCLIAHFTLRDPPMMMLDSSVILIMATTEFVULGNVSAIRTRVIALATISVIGLKI 240
QY 240 VQALIOSVKKLADVWVLTFCVSPFALIGLOFMGILRKCYRNFALNTGNSVADGL 299
Db 241 VQALIOSVKKLADVWVLTFCVSPFALIGLOFMGILRKCYRNFALNTGNSVADGL 300
QY 300 VESIEDLVLDPEBNYLKNGTSDVLLCGNSDAGTCPEGYRCLKAGENDHGYTSDSPA 359
Db 301 VVNSLDVYLVNDPANYLLKNGTIDVLLCGNSDAGTCPEGYRCLKAGENDHGYTSDSPA 360
QY 360 VAFALFLRLMTODCWERLYQOTLRSAKTYMIFMLVTFLSGYLVNLLAVVMAVEEQ 419
Db 361 VAFALFLRLMTODCWERLYQOTLRSAKTYMIFMLVTFLSGYLVNLLAVVMAVEEQ 420
QY 420 NOATTAEFEKERRFOEAMEMLKKEHEALTIRGVDTVSRSLSEMSPLAVNSHERSKR 479
Db 421 NOATTAEFEKERRFOEAMEMLKKEHEALTIRGVDTVSRSLSEMSPLAVNHERSKR 480
QY 480 KEMSSGTECEGDRLPKSDSEDPAMNHLSTRLGSRYSMKRPSRSGSIPTFRRRDLGS 539
Db 481 KRLSSGTEDGDDRLPKSDSEDPALNQLSLTHGLSRYSMKRPSRSGSIPTFRRRDLGS 540
QY 540 EADFPADDENSTIGSEBSHRTSLVWPPLRISAOQPSFGTSAFGHALHKKNSYDCCNG 599
Db 541 EADFPADDENSTIGSEBSHRTSLVWPPLRISAOQPSFGTSAFGHALHKKNSYDCCNG 600
QY 600 VVSLIAGDPEATSGSHLLRPVMLEHPDPTTPSEBPGPOMLTSOAPCVUGFEEBGR 659
Db 601 VVSLIAGDPEATSGSHLLRPVMLEHPDPTTPSEBPGPOMLTSOAPCVUGFEEBGR 660
QY 660 QRALSAVSILTSALTELESRRKCPCCWNRQLAQRVLIWECCLMMSIKQVGLVMDPPT 719
Db 661 QRALSAVSILTSALTELESRRKCPCCWNRQLAQRVLIWECCLMMSIKQVGLVMDPPT 720
QY 720 DLTITMCIVLNTLFPALBHYNMTSEPEMLQVGNLVFTGIFPAENTFKIADPYYYFQ 779
Db 721 DLTITMCIVLNTLFPALBHYNMTSEPEMLQVGNLVFTGIFPAENTFKIADPYYYFQ 780
QY 780 GNNIFDSIIIVLSLMEIGLSRMSNLVSRSPELNVPLAKSNPTLNTLIKINGSVGL 839
Db 781 GNNIFDSIIIVLSLMEIGLSRMSNLVSRSPELNVPLAKSNPTLNTLIKINGSVGL 840
QY 840 GNLTLVLAIIYFIFAVVGMQLFGKNYSELRLD--SDSGILPRWMMDFHAFLIIRILCG 897
Db 841 GNLTLVLAIIYFIFAVVGMQLFGKNYSELRLD--SDSGILPRWMMDFHAFLIIRILCG 900
QY 898 EWIETMDOMEVSGSGLCLVFLVLMVIGNLVLMFLALLISFSADNLTADEDEKN 957
Db 901 EWIETMDOMEVSGSGLCLVFLVLMVIGNLVLMFLALLISFSADNLTADEDEKN 960
QY 958 NQQLALARIQGLRVRKKTMDPCCGLRORPORALAAAOQLPSCATPVSPPPEPE 1017
Db 961 NQQLALARIQGLRVRKKTMDPCCGLRORPORALAAAOQLPSCATPVSPPPEPE 1020
QY 1018 KVPTRKREFECEGPOGQTIPGDEPVCVPIAVALSEPTDQOEDEENSLGTEBESSKOE 1077
Db 1021 KVPTRKREFECEGPOGQTIPGDEPVCVPIAVALSEPTDQOEDEENSLGTEBESSKOE 1080
QY 1078 SQVVSQGEAPDPDSRTWSQVSNVSSSEASASQADMFOQWKAEPQACGCTPEBSCSE 1137
Db 1081 SQVVSQGEAPDPDSRTWSQVSNVSSSEASASQADMFOQWKAEPQACGCTPEBSCSE 1140
QY 1138 GSTADMTYATLRLGIDPLGQDVVDPRDCFTGECVRCRCCANVTQOAPGVMMRLRKC 1197
Db 1141 GSTADMTYATLRLGIDPLGQDVVDPRDCFTGECVRCRCCANVTQOAPGVMMRLRKC 1200
QY 1198 YHIVHSWFETFIIFMILSSGALAFEDIVYEERKTIYVLEEVADKMFTYVFLVEMLMK 1257
Db 1201 YHIVHSWFETFIIFMILSSGALAFEDIVYEERKTIYVLEEVADKMFTYVFLVEMLMK 1260

QY 1258 VAYGFKYFTNAMCKDLIVDVSLVSLVANTLGFAMGPTKSLRTTLRALRPLRLSRFE 1317
DB 1261 VAYGFKYFTNAMCKDLIVDVSLVSLVANTLGFAMGPTKSLRTTLRALRPLRLSRFE 1320
QY 1318 GMRVYVNVLVGAIPSIIMNVLLVCLIFMLIFSIIMGNLPAKRGRCINOTEGDLPLANTYIV 1377
DB 1321 GMRVYVNVLVGAIPSIIMNVLLVCLIFMLIFSIIMGNLPAKRGRCINOTEGDLPLANTYIV 1380
QY 1378 NNSQCSLSNLJGELVMTKVVNFEDNVAGYLLALQVATFKGMDIMYAAVDSRGYEQP 1437
DB 1381 NNSQCSLSNLJGELVMTKVVNFEDNVAGYLLALQVATFKGMDIMYAAVDSRGYEQP 1440
QY 1438 QMEIVNLVYIFVFIITIGSPPTLNLFTGVIIIDNNQOKKLLGGODIMTEEQKKYNYAM 1497
DB 1441 QMEDNLVYIFVFIITIGSPPTLNLFTGVIIIDNNQOKKLLGGODIMTEEQKKYNYAM 1500
QY 1498 KKLGSKKPOKPIPRPLNKYOGFIDIVTKOAFDVTIMFLCLNMVYMMVETDDQSPKIN 1557
DB 1501 KKLGSKKPOKPIPRPLNKYOGFIDIVTKOAFDVTIMFLCLNMVYMMVETDDQSPKIN 1560
QY 1558 ILAKINLFLVAIFTEGCIIVKLAALRHYFTNSWNIJDFVNVILSIGTVLSDIIOKYEFS 1617
DB 1561 ILAKINLFLVAIFTEGCIIVKLAALRHYFTNSWNIJDFVNVILSIGTVLSDIIOKYEFS 1620
QY 1618 PTLFVIVILARIGRLILIRKAGKIRTLIFALMGLPALFNIGLLFLVMTFYSIRGMAN 1677
DB 1621 PTLFVIVILARIGRLILIRKAGKIRTLIFALMGLPALFNIGLLFLVMTFYSIRGMAN 1680
QY 1678 FAYVMEAGIDDMFOTFANSMLCLFOITTSAGWDGLSPILNTGPPYCDPTLPNSNGS 1737
DB 1681 FAYVMEAGIDDMFOTFANSMLCLFOITTSAGWDGLSPILNTGPPYCDPTLPNSNGS 1740
QY 1738 RGDGSPAVGILFPTTYIIISFLIVNMVYIILLENFSVAITEESTEPILSEDDFDMFYIWM 1797
DB 1741 RGDGSPAVGILFPTTYIIISFLIVNMVYIILLENFSVAITEESTEPILSEDDFDMFYIWM 1800
QY 1798 EKFDPEAOTFIYSVLSPADALSEPLRLAKNQISLIMDLPMVSGRIRHMDLFAFT 1857
DB 1801 EKFDPEAOTFIYSVLSPADALSEPLRLAKNQISLIMDLPMVSGRIRHMDLFAFT 1860
QY 1858 KRVLESSEGMALAKIOMEKEMAPNSKISYEPIITTLRKRKEEVSAMVIOQAPRRHLQ 1917
DB 1861 KRVLESSEGMALAKIOMEKEMAPNSKISYEPIITTLRKRKEEVSAMVIOQAPRRHLQ 1920
QY 1918 RSLKIASFLFRQA-GSGISEDAPEREGLIAYVNSEFSRPLGPPSSSSISSTISFPSPSY 1976
DB 1921 RSLKIASFLFRQA-GSGISEDAPEREGLIAYVNSEFSRPLGPPSSSSISSTISFPSPSY 1980
QY 1977 DSVTRATSDNLPVRASDYSRSEDLADFPSPDRRESIV 2015
DB 1981 DSVTRATSDNLPVRASDYSRSEDLADFPSPDRRESIV 2019

RESULT 22

AAR06584 standard; protein; 2020 AA.

AAR06584;

10-JAN-1991 (first entry)

Cardiac sodium channel.

Rat; arrhythmia.

Rattus rattus.

WO9009391-A.

23-AUG-1990.

13-FEB-1989; 89US-00310330.

XX 13-FEB-1989; 89US-00310330.
PR (ARCH-) ARCH DEV CORP.
XX Rogart RB;
XX WPI; 1990-275095/36.
DR N-PSDB; AA005831.
XX New rate cardiac sodium channel proteins - and associated DNA sequences,
PT polypeptide(s) and peptide(s) associated with proteins, useful as
PT antiarrhythmic and cardiotoxic drugs.
XX Disclosure; Fig 2; 65pp; English.
XX The sequence deduced from cDNA derived from 3 overlapping clones, pRH-
CC 1, pRH-23, and pRH-31. (Deposited as ATCC 67885, 67886, and 67887
CC resp.) The clones were isolated from a cDNA library in the lambda Zap
CC vector prep. from mRNA obtd. from newborn rat hearts using rat brain II
CC cDNA probe. The protein has diagnostic, therapeutic, and prognostic
CC applications
XX
SQ Sequence 2020 AA;
Query Match 92.7%; Score 9719; DB 2; Length 2020;
Best local similarity 93.1%; Pred. No. 0;
Matches 1879; Conservative 44; Mismatches 92; Indels 4; Gaps 3;
QY 1 MANLPLRGTSFRRFRESLIAIEKMAEKQAR-GSTYLOESREGLPDEAPRPOLDQ 59
DB 1 MANLPLRGTSFRRFRESLIAIEKMAEKQAR-GSTYLOESREGLPDEAPRPOLDQ 60
QY 60 ASKLPDLXGNPPELIGEPLEDDLPYSTOKTFIVLNKGTIFRESATNALVLSFPH 119
DB 61 ASKLPDLXGNPPELIGEPLEDDLPYSTOKTFIVLNKGTIFRESATNALVLSFPH 120
QY 120 JRRRAVILVHSLFNMILMCTIILNCVFMQOHPPTKTYVEYFTAIYTFESIVKLAR 179
DB 121 JRRRAVILVHSLFNMILMCTIILNCVFMQOHPPTKTYVEYFTAIYTFESIVKLAR 180
QY 180 GFCLHAFTFLRDPNMWLDPSVYIIMAYTTEFVDLGNVSLRTPFRVLRALKTISVIGLKI 239
DB 181 GFCLHAFTFLRDPNMWLDPSVYIIMAYTTEFVDLGNVSLRTPFRVLRALKTISVIGLKI 240
QY 240 VGALIOGVKGLADVMVLTVPCLSVFALIGLOLPMGNLRHCVNRFTELNGNSVEADGL 299
DB 241 VGALIOGVKGLADVMVLTVPCLSVFALIGLOLPMGNLRHCVNRFTELNGNSVEADGL 300
QY 300 VMSLIDYLDSPENYLLKNGTSDVLLCGNSDAGTCEGYRCLAKGENPDHGYTSPDFA 359
DB 301 VMSLIDYLDSPENYLLKNGTSDVLLCGNSDAGTCEGYRCLAKGENPDHGYTSPDFA 360
QY 360 WAPLALFRLMTQDCMERLYOQTLSAGKIYIMFMLVIFLGSFYLVNLIILAVMAYBEQ 419
DB 361 WAPLALFRLMTQDCMERLYOQTLSAGKIYIMFMLVIFLGSFYLVNLIILAVMAYBEQ 420
QY 420 NOATIAETEEKERFOEMEMWLKKEHEALTRIGDVTYSRSSLEMSPLAPVNSHRSRKR 479
DB 421 NOATIAETEEKERFOEMEMWLKKEHEALTRIGDVTYSRSSLEMSPLAPVNSHRSRKR 480
QY 480 KRMSSGTEECGEDRLPKSDSEDDGRANMHLISLRTGLSRTSMKPRSSRGSITFPERRDLGS 539
DB 481 KRMSSGTEECGEDRLPKSDSEDDGRANMHLISLRTGLSRTSMKPRSSRGSITFPERRDLGS 540
QY 540 EADPADDENSTAGESSEHSRTSLVWPPLRSTSAQGPSPTGSAPGHALHKGKNSVTDCNG 599
DB 541 EADPADDENSTAGESSEHSRTSLVWPPLRSTSAQGPSPTGSAPGHALHKGKNSVTDCNG 600
QY 600 VVSLIAGDPPATSPGSHILARPVNLHHPDPTTSSERGGQMLTSQAPCVDGFEPPGAR 659
DB 601 VVSLIAGDPPATSPGSHILARPVNLHHPDPTTSSERGGQMLTSQAPCVDGFEPPGAR 660

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Qy 660 QALSAVSVLTSALEELBSRHKCPCCWNRLLAQRVLIWECCLMMSIKQVGLVYMDPEPT 719
Db 661 QRALSAVSVLTSALEELBSRHKCPCCWNRFPQHLIWECCPLMMSIKQKXVFVMDPEFA 720
Qy 720 DLTITMCIVLNTLFNALHYNMTSFEEMLOVGNLVFTGIFPAENTFKIALDPYTYRQO 779
Db 721 DLTITMCIVLNTLFNALHYNMTSFEEMLOVGNLVFTGIFPAENTFKIALDPYTYRQO 780
Qy 780 GNNIFDSITVILSLMEIGLSRMSNLSVLSRPFLLVFPFLAKSWPNTLTIKIGNSVAL 839
Db 781 GNNIFDSITVILSLMEIGLSRMSNLSVLSRPFLLVFPFLAKSWPNTLTIKIGNSVAL 840
Qy 840 GNLTLVLAIVFIFAVVGWQLFGKNYSBLRD--SDSGLLPRWMDPFHAPLIRILCG 897
Db 841 GNLTLVLAIVFIFAVVGWQLFGKNYSBLRHRSISGGLPRWMDPFHAPLIRILCG 900
Qy 898 EWIETMDOMEVSGSLCLVFLVMTIGNLVNLPLALLISFSADNLTAPDEDEMN 957
Db 901 EWIETMDOMEVSGSLCLVFLVMTIGNLVNLPLALLISFSADNLTAPDEDEMN 960
Qy 958 NLQIALARIQGLRFRKRTMDRCGGLRORPQRAALAAQGLPSCATPYSPPEPTE 1017
Db 961 NLQIALARIQGLRFRKRTMDRCGGLRORPQRAALAAQGLPSCATPYSPPEPTE 1020
Qy 1018 KVPTRKRETRFEGRPOGQGTGDEPEVCVPIAVALSDTDDEBDEENSLGTEESSKOE 1077
Db 1021 KVPTRKRETRFEGRPOGQGTGDEPEVCVPIAVALSDTDDEBDEENSLGTEESSKOE 1080
Qy 1078 SQPVSGGEPAPDSHTWSQVATASSEBASASQADMWQWKAEPQACGCTPEPDSCE 1137
Db 1081 SQPVSGGHEPQEPAPMSQVSETTSSEAGASTSQADMWQEQTEPQACGCTPEPDSYSE 1140
Qy 1138 GSTADMTYTAELBEQIPDLGQDVKRPBDCFTGCGRCRRPCCAVDTTQAGKMWALRTKC 1197
Db 1141 GSTADMTYTAELBEQIPDLGQDVKRPBDCFTGCGRCRRPCCAVDTTQAGKMWALRTKC 1200
Qy 1198 YHIVHSWFEFTIIMILSSGALAFEDIYLEERKTIKYLEYADKMTYVYVLEMLTKM 1257
Db 1201 YHIVHSWFEFTIIMILSSGALAFEDIYLEERKTIKYLEYADKMTYVYVLEMLTKM 1260
Qy 1258 VAYGFKKYFTNAMCWLDFLIYDVSLVSLVANTLGAEMGPIKSLTTLRALRPLRALSPE 1317
Db 1261 VAYGFKKYFTNAMCWLDFLIYDVSLVSLVANTLGAEMGPIKSLTTLRALRPLRALSPE 1320
Qy 1318 GMRVYVNALVGAIPESINAVLVCLIFPLIFSGIMGNVPAKGRGRCINOTEGDPLANTYIV 1377
Db 1321 GMRVYVNALVGAIPESINAVLVCLIFPLIFSGIMGNVPAKGRGRCINOTEGDPLANTYIV 1380
Qy 1378 NNSKQCESLNTLGEIYMTKVKVKNPDNVAGYALALQVATPFKGMMDIMYAAVDSRGEYEROP 1437
Db 1381 NNSKQCESFNVTGELIYMTKVKVKNPDNVAGYALALQVATPFKGMMDIMYAAVDSRGEYEROP 1440
Qy 1438 QMEVNLVMTYIFVFIIFGSPFTLNLFTGVIIIDNENQOKKGLGQODIEMTESOKKYANAM 1497
Db 1441 QMEBNLVMTYIFVFIIFGSPFTLNLFTGVIIIVENQOKKGLGQODIEMTESOKKYANAM 1500
Qy 1498 KKLGSKKPQKPIPREPLANKYQGFIPDIVTKQAFDVTIMFLICLNMVMTMVEETDOSPCKIN 1557
Db 1501 KKLGSKKPQKPIPREPLANKYQGFIPDIVTKQAFDVTIMFLICLNMVMTMVEETDOSPCKIN 1560
Qy 1558 ILAKINLFFVAIPFGECEIVKLAAALRHYFTNSWNI FDEVVYVLSIVGVLSIIIOKYPFS 1617
Db 1561 ILAKINLFFVAIPFGECEIVKLAAALRHYFTNSWNI FDEVVYVLSIVGVLSIIIOKYPFS 1620
Qy 1618 PTLFRVIRIARIGRLIRLIRGAKGIRTLIFALMMSLPALFNIGLLFLVMEFYSIFGMAN 1677
Db 1621 PTLFRVIRIARIGRLIRLIRGAKGIRTLIFALMMSLPALFNIGLLFLVMEFYSIFGMAN 1680
Qy 1678 FAYYKMEGIDMMEFQTFPANSMLCLFOITTSAGMDGLSLPLANTGPYCPDPLNPSNGS 1737
Db 1681 FAYYKMEGIDMMEFQTFPANSMLCLFOITTSAGMDGLSLPLANTGPYCPDPLNPSNGS 1740
Qy 1738 RGDGSPAVGILFFTYTIIISFLIVNNYIAIILENFVATBEESTEPLESDDFMFEYIWM 1797

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Db 1741 RNCGSPAVGILFFTYTIIISFLIVNNYIAIILENFVATBEESTEPLESDDFMFEYIWM 1800
Qy 1798 EKPDPEAQOFIEYISVLSFPADALSEPLAIAPNOISLNMPLPMVSGRIRHMDILFAPT 1857
Db 1801 EKPDPEAQOFIEYISVLSFPADALSEPLAIAPNOISLNMPLPMVSGRIRHMDILFAPT 1860
Qy 1858 KRVLGSEGMALKIOMEKEMANPSKISYEPIITTLRRKHGEVSAMVIORAFRRHLQ 1917
Db 1861 KRVLGSEGMALKIOMEKEMANPSKISYEPIITTLRRKHGEVSATVIORAFRRHLQ 1920
Qy 1918 RSLKRAEFLFROQA--GSGLSEEDAPEREGLIAYVNSENFSRPLGPPSSSISSTSPPSY 1976
Db 1921 RSVKRAEFLFROQA--GSGLSEEDAPEREGLIAYVNSENFSRRAAPLSSSISSTSPPSY 1980
Qy 1977 DSVTRATSDNIQVRGSDYSHSEDLADPPSPDRDRRESIV 2015
Db 1981 DSVTRATSDNLPVRASDYSRSEDLADPPSPDRDRRESIV 2019

RESULT 23
AAU19518
ID AAU19518 standard; protein; 1603 AA.
XX
AC AAU19518;
XX
DT 04-DEC-2001 (first entry)
XX
DE Human diagnostic and therapeutic polypeptide (DITMP) #104.
XX
KW Human; receptor; diagnostic; therapeutic; gene therapy; vaccine;
KW cell proliferative disorder; Crohn's disease; lymphoma; leukaemia;
KW acquired immune deficiency syndrome; AIDS; autoimmune disorder;
KW respiratory disorder.
XX
OS Homo sapiens.
XX
PN MO200162927-A2.
PD 30-AUG-2001.
XX
PF 21-FEB-2001; 2001WO-US006059.
XX
PR 24-FEB-2000; 2000US-0184693P.
PR 24-FEB-2000; 2000US-0184697P.
PR 24-FEB-2000; 2000US-0184698P.
PR 24-FEB-2000; 2000US-0184768P.
PR 24-FEB-2000; 2000US-0184769P.
PR 24-FEB-2000; 2000US-0184770P.
PR 24-FEB-2000; 2000US-0184771P.
PR 24-FEB-2000; 2000US-0184772P.
PR 24-FEB-2000; 2000US-0184773P.
PR 24-FEB-2000; 2000US-0184774P.
PR 24-FEB-2000; 2000US-0184776P.
PR 24-FEB-2000; 2000US-0184777P.
PR 24-FEB-2000; 2000US-0184779P.
PR 24-FEB-2000; 2000US-0184797P.
PR 24-FEB-2000; 2000US-0184813P.
PR 24-FEB-2000; 2000US-0184837P.
PR 24-FEB-2000; 2000US-0184841P.
PR 24-FEB-2000; 2000US-0185213P.
PR 24-FEB-2000; 2000US-0185216P.
PR 24-FEB-2000; 2000US-0185219P.
PR 12-MAY-2000; 2000US-0203785P.
PR 15-MAY-2000; 2000US-0204226P.
PR 16-MAY-2000; 2000US-0204525P.
PR 16-MAY-2000; 2000US-0204821P.
PR 16-MAY-2000; 2000US-0204908P.
PR 16-MAY-2000; 2000US-0205232P.
PR 17-MAY-2000; 2000US-0204815P.
PR 17-MAY-2000; 2000US-0204863P.
PR 17-MAY-2000; 2000US-0205221P.
PR 17-MAY-2000; 2000US-0205285P.
PR 17-MAY-2000; 2000US-0205286P.
PR 17-MAY-2000; 2000US-0205287P.

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DB 1431 IVNNKSDCKIONSTGSGFFWNVNKNFNDVAMGYLALLQVATPKGMMWIMYAVDSREVNM 1490
 QY 1436 OPQWENLYMYTYFYFIITFGSFTLNLPIGYIINPNQKKKGODIEMTEBOKRYN 1495
 DB 1491 QPKMDNVMYMYFYFIITFGSFTLNLPIGYIINPNQKKKGODIEMTEBOKRYN 1550
 QY 1496 AMKKLGSKKKPKQKIPRPLNKYOGFIEDIVTKQAFDVITIMFLICLNMV 1542
 DB 1551 AMKKLGSKKKPKQKIPRPLNKYOGFIEDIVTKQAFDVITIMFLICLNMV 1597

RESULT 24

ADP79541

ID ADP79541 standard; protein; 2000 AA.

AC ADP79541;

DT 04-NOV-2004 (first entry)

DE Human sodium III channel splice variant (hna11118).

KM Human; sodium III channel; hna11118; splice variant; antiarrhythmic;

KN analgesic.

OS Homo sapiens.

PN MO2004050857-A2.

PD 17-JUN-2004.

PF 04-DEC-2003; 2003WO-US038796.

PR 04-DEC-2002; 2002US-0431794P.

PX (EURO-) EUROCELLTQUE SA.

PY Kammesheidt A, Hodges D;

PX MPI; 2004-450725/42.

DR N-P8DB; ADP79540.

PT New human sodium III channel, useful in treating cardiac arrhythmias,
 PT herpes virus infection, diabetes mellitus, or vasculitis.

PS Claim 9; SEQ ID NO 2; 101pp; English.

CC The present sequence is that of a novel splice variant of the human
 CC sodium III channel alpha subunit, denoted hna11118. The cDNA was cloned
 CC by RT-PCR from human embryonic brain total RNA using human Natrii specific
 CC primers ADP79546-ADP79547. The hna11118 sequence contains an additional
 CC 49 amino acid residues that do not appear in a previously reported splice
 CC Natrii ADP79543, and also differs from a previously reported splice
 CC variant of human sodium channel alpha subunit ADP79545 by 12 amino acids
 CC out of 2000. Transient transfection of HEK293 cells by hna11118 resulted
 CC in expression of functional sodium channels. The invention provides:
 CC hna11118 proteins and their fragments and derivatives; hna11118-encoding
 CC nucleic acids and their fragments, including primers, probes, and
 CC regulatory sequences; hna11118-specific antibodies; and methods of using
 CC these materials to detect the presence of hna11118 proteins or nucleic
 CC acids. The invention also provides an assay method for screening to
 CC identify selective modulators of hna11118 expression or activity. These
 CC may be useful for treatment of conditions associated with sodium channel
 CC over- or under-expression, e.g. for treatment of cardiac arrhythmias,
 CC neuronal disorders, nociceptive pain-related diseases and neuropathic
 CC pain-related diseases, e.g. pain from peripheral nerve trauma, herpes
 CC virus infection, diabetes mellitus, causalgia, plexus avulsion, neuroma,
 CC limb amputation or vasculitis.

SQ Sequence 2000 AA;

Query Match 61.1%; Score 6406.5; DB 8; Length 2000;
 Best Local Similarity 62.8%; Pred. No. 0;
 Matches 1297; Conservative 232; Mismatches 366; Indels 171; Gaps 28;

QY 5 LLPRGTSFRRFTRESLAIEKRMALKEQARSGTTLQESRBLPEEAPRPQDLQASKL 64
 DB 6 LVPGPBSFRLPTRRSALAIERBAEKAKEKKPKKEQDN-----DDEKKKPNNDLEAGNKL 61
 QY 65 PDLGNPPOBLIGEPLEDDPPYSTOKTPIVANKKTIIPRSANALVYLSDFHPIRRA 124
 DB 62 PIYVDIDPEWVSEPLEDDDYINKTFIIVNKGKAIIPRSATSALYILFPLNVRKIA 121
 QY 125 VKILVHSLFNNLIMCTIITNCVPAQHPDPWTKVETTFATYFESLVILAGFCLH 184
 DB 122 IKILVHSLFNNLIMCTIITNCVPAQHPDPWTKVETTFATYFESLVILAGFCLH 181
 QY 185 APTFLRDPNNMLDPSVIIMAYTTEPVDIGVNSALRTFVPLAKTISYISGLKTVGALI 244
 DB 182 DPTFLRDPNNMLDPSVIIMAYTTEPVDIGVNSALRTFVPLAKTISYISGLKTVGALI 241
 QY 245^QSVKKGLADVMVLYVECLSVFALIGLQFMGNLRHKVCR-----NFTAL----- 287
 DB 242 QSVKKGLADVMVLYVECLSVFALIGLQFMGNLRHKVCR-----NFTAL----- 287
 QY 288 NGTNGSVADGLWESLDLYSDPENYLLKNGTSVLLCGSSSDAGTCEPGRCLAKGEN 347
 DB 302 NGTFVNVMTSTFNKMD---YIGDSSHFYVLDGQDPLICGSGSDAGQCEGYICVAGRN 358
 QY 348 PDHGYTSPDSFAMAFIALFRLMTODCWERLYQOOLRSAGKLYMIFPMVIFAGSPYVNL 407
 DB 359 PNYGTTSDITSMAFSLFRLMTODYENLQULIRAKGKTYMIFVLYIFLGSFYVNL 418
 QY 408 ILAVVAMAYEONQATTAEKEKRRFOAMEMLKKEHALT-----IRGV 453
 DB 419 ILAVVAMAYEONQATTAEKEKRRFOAMEMLKKEHALT-----IRGV 453
 QY 454 DTVSRSSLEMSPLAPVNSHERSKRRKMS-----SGTECEGDELPKDSDEGPAAHML 509
 DB 479 GELLESESEAKLSKSAKEWRNRKRRRREHLEGNNGKGRDSEPKSESSESVRSFL 538
 QY 510 SLTRGLSRTSMKP-----RSRGSIFTFRRR--DLGEADPADEN 548
 DB 539 FSDQGNRLTSDKKCSPIQSLSTRGSLFSRRRSKTSISFSGRAKGVSENDPADEN 598
 QY 549 STAGESESHRTSLVP--WPLRRTSAQGPSPGT--SAPGHALHGKNSVDCNGVSLG 605
 DB 599 STPDESSESRDLSLFPVHRHGERRNSVNSQAAMSRMVGLPANGMHSITVDCNGVSLG 658
 QY 606 AGDPBATSQSHLRPVMLHEHPPTTPSEBGPQMLTSAQPCVDGEEBGAQRALSA 665
 DB 659 -GPAALTSPTQL-----PPEGTT--TETEVRRRLSSYQISMEMLEDSGRORAVSI 708
 QY 666 VSVLTSLMEERESRHKCPQCNMLAQRVLIWECCPLMMSIKQGVKLVMDPFDLTITM 725
 DB 709 ASILTNTEBESRQKCPQWYRPAVFLWDCDDALVKVHLVNLVMDPFDLALITI 768
 QY 726 CIVANTLPMALHNMTSEFEEMLOVGNLVTGIFTAEMTKIALDPYVYFOQGMNIFD 785
 DB 769^ACIVANTLPMALHNMTSEFEEMLOVGNLVTGIFTAEMTKIALDPYVYFOQGMNIFD 828
 QY 786 SIIVLSIMEIGLSRMSNLVLRSGFRLIRVFKLAKSWPTLNTLIKIIGNSVAGLGNLTV 845
 DB 829 GIIVLSIMEIGLSRMSNLVLRSGFRLIRVFKLAKSWPTLNTLIKIIGNSVAGLGNLTV 888
 QY 846 LAIVVFPAVVGMLPGKNYSE--LRDSGSLPRMNMDFHAFILIFRILICGEWRTM 903
 DB 889 LAIVVFPAVVGMLPGKNYSE--LRDSGSLPRMNMDFHAFILIFRILICGEWRTM 948
 QY 904 WDCEVSGQSICLVFLVAVVIGNLVVLNTLALILSSFSADNLTAPDEDEMMNLQVAL 963
 DB 949 WDCEVAGQTMCLVFLVAVVIGNLVVLNTLALILSSFSADNLTAPDEDEMMNLQVAL 1008
 QY 964 ARIGRLRFVKKRTTWDFCCGLLRPQKPAALAAQGLPSCIATPIYSEPPETKEVPTTR 1023
 DB 1009 GRMGKIDIVYNNKRR--CFQKAPFRKRVIEIHGKNIDSCMSNNTG-----IEISKEL 1061

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QY 1024 KETPEEBOGPQGTSPDPE-----PVCVPIAVALSEDTDDQEDSEN 1065
Db 1062 NYLIDGNNTTSGVGSSVEKKYIDENDYMSFINNPSTLVTVPLAVGSDPE----- 1113
QY 1066 SLGTEESSKOESQOPVSGAPAPDSRTWSQVSATASAEASASAOAMROQMKAEPQAP 1125
Db 1114 NLNTEFESESELE-----EKEKLNANSS----- 1138
QY 1126 GCGETPEDSCSEGSTAD--MTNTAELFQIPDLGDVDKPEDCFTEGCVRCPCCAVDTT 1183
Db 1139 -----SEGSTVDVPLPREGQAEETPE--EDLK-PEACFTEGCIKKFPQCVSTE 1185
QY 1184 QAPGVWRLRKTCTCHIEHSEHFEFIIIMLLSSGALAFEDYIEBKRTTKVLEEVADK 1243
Db 1186 EGKGIMWNLRKTCYSIEHMFETFIIVFMILLSSGALAFEDYIEBKRTTKVLEEVADK 1245
QY 1244 MFTYFVLEMLLKNVAYGFKKFTNAMCMLDFLIDVSLVSLVANTLGLFAEMGPIKSLRT 1303
Db 1246 VFTYFIEMLLKNVAYGFGQYFTNAMCMLDFLIDVSLVSLVANTLGLSELGAIKSLRT 1305
QY 1304 LRALRPLALSRFEGMRYVNVAVGALPSIMNVLLVCLIPWLISIMGNVLPAGKRGCT 1363
Db 1306 LRALRPLALSRFEGMRYVNVAVGALPSIMNVLLVCLIPWLISIMGNVLPAGKRGCT 1365
QY 1364 NOTBEDLPLNTIYVNNKSCESLNTGELVYTKVNVNPDNVAGVLLALQVATEKGMMDI 1423
Db 1366 NMTTGNM--FDISDVNNLSDCCQLG--KQARMKNVNVNPDNVAGVLLALQVATEKGMMDI 1422
QY 1424 MYAAVDSRGVEQPOMEVNYLYVYIFVIFIIIFGSEFTLNLFGVLIIDNFNOQKKLGGSD 1483
Db 1423 MYAAVDSRDVQLQPYEENLYNYLFFVIFIIIFGSEFTLNLFGVLIIDNFNOQKKLGGSD 1482
QY 1484 IFMTBEQKYYNAMKKLGSKKFQKPIPRELNTKYQPIPIPIVYTKQAPDVTIMEFLILANVT 1543
Db 1483 IFMTBEQKYYNAMKKLGSKKFQKPIPRELNTKYQPIPIPIVYTKQAPDVTIMEFLILANVT 1542
QY 1544 MMVETDDOSPEKINILAKINILFVAIFGECVCLKALRHVYFTSMNIPDVVVILSLV 1603
Db 1543 MMVETDDOSKWTLVLSRLNLFVLFGEFELRLVLSLHYYFTIGMNIPEVVVILSLV 1602
QY 1604 GTVLSDIIOKYPFSPFLTFVIRIARIGRIILIRAKAGIRTLIFALMMSLPALFNIGILL 1663
Db 1603 GMFLAMEIEKTVSPTLPRVIRIARIGRIILIRAKAGIRTLIFALMMSLPALFNIGILL 1662
QY 1664 FLVMTYISIFGMANFAYVKEAGIDMNFQTPANSMCLPFIOTTSAGMDGLSLPLNTG 1723
Db 1663 FLVMTYIYAFGMSNPAFYVKEAGIDMNFQTPANSMCLPFIOTTSAGMDGLSLPLNTG 1722
QY 1724 PRCYCP-TPPNNGSRGDCGSPAVGILFTTYIIISPLIVNMYTAIILENFSVATEEST 1782
Db 1723 PRCYCPDPTIHPGSSVYKDCGNPSVGIFFVVSYIIISPLIVNMYTAIILENFSVATEEST 1782
QY 1783 EPLSDDDFPMFEIWEKEPPEATQFIYEVLSDPDALSEPLIAKPNQISLINDLPMV 1842
Db 1783 EPLSDDDFPMFEIWEKEPPEATQFIYEVLSDPDALSEPLIAKPNQISLINDLPMV 1842
QY 1843 SCDRIHCDMILFAFTKRVLVGSEGMALKIOMEKFMANPSKISYEPIITTLRKHEEV 1902
Db 1843 SCDRIHCDMILFAFTKRVLVGSEGMALKIOMEKFMANPSKISYEPIITTLRKHEEV 1902
QY 1903 SAMVYQARFRLHLRSLKGLASFLPRQOQSGSGLSEDAERBGLLAVYVSENFSPRLGPP 1962
Db 1903 SAAIIQORNFRCYLLKQRLKNISSNYKEAIKG--RIDLPKIDOMIIDKLNGNST---PE 1956
QY 1963 SSSSISSTSPSPSYDSVTRATSDNIQ 1988
Db 1957 KTDGSSSTTSPSPSYDSVTRATSDNIQ 1982

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AC AAB99676.i
XX
XX 04-SEP-2001 (first entry)
DT
XX
XX Human adult form of SCN2A protein sequence SEQ ID NO:35.
DE
XX
XX Human; epilepsy; chromosome 2; SCN1A; SCN2A; SCN3A; identification;
KW diagnosis; mutation; chromosome 2q23-q31; neurological disorder;
KM anticonvulsant; neuroprotective.
XX
XX Homo sapiens.
OS
XX
XX WC020138564-A2.
XX
XX
XX 31-MAY-2001.
XX
XX 24-NOV-2000; 2000WO-CA001404.
XX
XX 26-NOV-1999; 99US-0167623P.
XX
XX (UTMC-) UNIV MCGILL.
PA
XX
XX Rouleau GA, Latreiniere RG, Rochefort D, Cossette P, Rasedale D;
PI N-PSDB; AAH55793.
DR
XX
XX WPI; 2001-355945/37.
PT
XX
XX Determining a predisposition to epilepsy and/or development of epilepsy
PT comprises determining the genotype of SCN1A, SCN2A and/or SCN3A, or a DNA
variant, equivalent, or mutation which shows a linkage disequilibrium.
PS Disclosure: Page 123-130; 268pp; English.
XX
XX The present invention describes a method (M1) of determining an
XX individual's predisposition to epilepsy and/or development of epilepsy,
XX as well as predicting the individual's response to medication. The method
XX comprises determining the genotype of at least one gene selected from
XX SCN1A, SCN2A or SCN3A, or a DNA variant, equivalent, or mutation which
XX shows a linkage disequilibrium. SCN1A, SCN2A and SCN3A are all sodium
XX channel genes located on chromosome 2. The idiopathic generalised
XX epilepsy (IGE) gene is more specifically localised on chromosome 2q23-
XX q31. Compounds identified as modulators of the biological activity of
XX SCN1A, SCN2A or SCN3A proteins or genes, are useful for treating epilepsy
XX or other neurological disorders. They have anticonvulsant and
XX neuroprotective activities. AAH55763 to AAH56164 and AAB99674 to AAB99679
XX represent SCN1A, SCN2A, and SCN3A cDNAs, gene fragments, PCR primers,
XX oligonucleotides and proteins given in the exemplification of the present
XX invention
XX
XX Sequence 2005 AA:
SQ
Query Match 61.0%; Score 6394.5; DB 4; Length 2005;
Best Local Similarity 62.2%; Pred. No. 0;
Matches 1301; Conservative 231; Mismatches 360; Indels 201; Gaps 33;
QY 5 LLPGTSEFRFBTRESLAIEKMAEKOARSTTLQESREGPEPEARPOLDLOASKTL 64
Db 6 LVPEPDSFRFBTRESLAIEKMAEKOARSTTLQESREGPEPEARPOLDLOASKTL 62
QY 65 PDLVGNPQOEILGEPLDLDPEFYSTOKTFIYLVNGKTIIFRSATNALVYVSPFHDIRRA 124
Db 63 PFIYDIDPEVWVSLDLDPEYIYINKKTFIYLVNGKAIKRSATPALYILPFPNIRKLA 122
QY 125 VKILVHSLFNNLMIMCTIITNCVFNMAQNDPPWTYKVEYTFPAIYTFESLVKILARGFLH 184
Db 123 IKILVHSLFNNLMIMCTIITNCVFNMTSNPDMTKVETFTGIIYFESLILKILARGFLE 182
QY 185 AFTFLRDPMNMLDPSVITIMATYTFEVDLGNYSALRTFPAVLAALKTISYISGLKTIYVGLI 244
Db 183 DFTFLRDPMNMLDPSVITIMATYTFEVDLGNYSALRTFPAVLAALKTISYISGLKTIYVGLI 242
QY 245 QSVKILADVMVLVYFCLSVFALIGLQFMGNLRHKCV-----NFTLANGTNGSV 294

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RESULT 25
 AAB99676
 ID AAB99676 standard; protein; 2005 AA.
 XX

Db 243 OSVKKLSDMVILITFLCLSVFALIGLQLFMGNLKNKCLQWPDNSSEFINITSF-- -FNNSL 300
 QY 295 EADGLWME-----SIDLYLSPDENLKNKTSVDLCCNSSPAGCCPGYRCLKGENP 348
 Db 301 DNGGTFENRTVSIENNDEYIEDSHFYFLEGQNDALLCNSSDAGCCPGYTCVKGGRNP 360
 QY 349 DHGYSFSDSPAFALFLPLMTQDCWERTLYOQTLRSAGKIYMFPLVYFLSGFYLVNLI 408
 Db 361 NNGYSFDFRFSNAFLSLPLMTQDPWENLYQTLRPAAGKIYMFPLVYFLSGFYLVNLI 420
 QY 409 LAVVANAAYEONQATTAETBEKRFQEAEMEMLKKEHEALTIR-----GV 453
 Db 421 LAVVANAAYEONQATLEAEQKEAEFQOMLEQLKQOEBAQAAAAAASERDPFGAGGI 480
 QY 454 DTVSRSSLEMSPLAPVNSHE---RKRKRKRMSGGTEEGEDPLPDSDEDR----- 504
 Db 481 GVFSSESSVASKLSSSEKELKNRRKKKKQKEQSGEE- KNDRVLSSESDSIRRGFRF 539
 QY 505 -----AMNHLSTRGLSRTSMKPRSSRSIFTPRRR--DLGSEADPADENS 549
 Db 540 SLEGSRLTYEKRFSSPHQSLSTRGLSPRRNRASLTSFRGRADIGSENDPADENS 599
 QY 550 TAGESESHRTSLVP--WPLRRTSAGQSPSGTSA-PGHALHGKNSYDCCNGVSLGA 606
 Db 600 TEEDNDSRRDSLFPVPRHGERRRHSVQASRASRVLPILPMNGKMSAVDCNGVSLVG- 658
 QY 607 GDBEATSPGSHLLRPMLHPPTTTPSEBPGPQWLTQAPCVQDFEERGARQRLSAV 666
 Db 659 GSGTSLTSAQQL-----PGTTEETEI--RKRSSSYHVSMDLEDPTRORAMSTA 708
 QY 667 SVLTSLAELEESRHKPCPCMNRLAORYIWECCPLMMSIKQGVKLWVDPFDLTITMC 726
 Db 709 SIIITNMEELSESRQCPWCYKAFAMCIWDCKRWLVKHLVNLVMDPFDAIATTC 768
 QY 727 IVLNTLPMALBHYNMTSEFEEMLOVGNLVFTGIFTAEMTKIITADPYTYFOGNNIFDS 786
 Db 769 IYNTLTFMAHEHYPMTEGSSVLSVGNLVFTGIFTAEMTKIITADPYTYFOGNNIFDS 828
 QY 787 IIVIIISLMEGLSRNSNLVLSRFLRPFKLAKSPPTLNTLIKIGNSVGLAGNLTLYL 846
 Db 829 FIVSLISLMEGLANVEGLSVLRSFLLRPFKLAKSPPTLNTLIKIGNSVGLAGNLTLYL 888
 QY 847 AIIIVFIAVVGQOLFQKNYS--LRDSDGLPRHMDPFAPFLIIFRILGEMETMW 904
 Db 889 AIIIVFIAVVGQOLFQKSYKECVCKISNDCELPRIHMDPFHSLFIVYLCGEMETMW 948
 QY 905 DCMAYSGOSLCLVFLVNVNIGNLVNLFLALLSSFSADNLTAPDEDEMNNTQLALA 964
 Db 949 DCMAYAGQTMCLTFPMVWVIGNLVNLFLALLSSFSADNLTAPDEDEMNNTQLIAYG 1008
 QY 965 RIQRLGRFVYKRTTMDPCCGLRQRPQKPAAL-----AAQQLPSCIAPTYSPPEETE 1017
 Db 1009 RMQKQIDFVKRKIRF--IQKAFVKQKALDEIKPLEDLANKKOSCIS----- 1054
 QY 1018 KVPTRKETRFEEG-----OPGQGRP-----GDPE-PVCVPIA 1050
 Db 1055 -----NHTTETIGDNLKNGNGTTSIGSSVEKTYVDESDFWSPINNSPLTYVPIA 1108
 QY 1051 VAESDPTDOEBDEENSLGTEBESSKQESQVPSGPEAPDSTRWSQVATASSEABASAS 1110
 Db 1109 VQESDFE-----NINTEEFSSSDME-----ESKELNANTSSSEGST--- 1145
 QY 1111 QADWQOQKAEPQOAGCGETPEPDSSEGSTADMTTALLEOIPDLQGVQKPEDCFTG 1170
 Db 1146 -----VDIGAABEGQPE-----VEPEESLE-----PEACFTED 1174
 QY 1171 CVRRCPCCAVDTQAPGVKMWRLRKCYAIVHSHMPEPTIIMILLSSGALAFEDITYLE 1230
 Db 1175 CVRRKCCOISIEBKGKLMNMLRKTCYIVHNNMFETIVMILLSSGALAFEDITYLEQ 1234
 QY 1231 RKTIVLLEYADKMFYVLEMLKMWAVYGFKKYFTNACWLDPLIVDSLVSLVANTL 1290
 Db 1235 RKTIVLLEYADKMFYVLEMLKMWAVYGFYFTNACWLDPLIVDSLVSLVANTL 1294

QY 1291 GFABGPIKSLRTLRALRPLRALSREGKRVVNVNLVGAIPSIWNLVLCFLFMIIFSI 1350
 Db 1295 GTSBGALISLTLRLRLRPLRSRREGKRAVNVNLVGAIPSIWNLVLCFLFMIIFSI 1354
 QY 1351 GVNLPAGEKRCINQTEGDLPLNYTIVNNKSOCESY--NLGELYWYKRVNFDVVGAG 1407
 Db 1355 GVNLPAGEKRYHCINTTGGEM-FDVSVVNNYSCKALISBNQAR--KRVKVNFDVVGAG 1411
 QY 1408 YTALLQVATFKCMNDIMTAANDSRGEBOPQWENLYMYTYVYVIFIFSSFTLANLFTGV 1467
 Db 1412 YLSLLQVATFKCMNDIMTAANDSRGEBOPQWENLYMYTYVYVIFIFSSFTLANLFTGV 1471
 QY 1466 IIDNFQOKKKLGGODIFWTEBOKKYNNAMKLGSKKQKPIPRPLNKYQGFPOIVTQO 1527
 Db 1472 IIDNFQOKKKLGGODIFWTEBOKKYNNAMKLGSKKQKPIPRPLNKYQGFPOIVTQO 1531
 QY 1528 AFDVTIMPLICLNMVTVMTEDDQSEPEKINILAKINLFLVALFTGECIVKLAALRHYFT 1587
 Db 1532 VFDISIMILICLNMVTVMTEDDQSEPEKINILAKINLFLVALFTGECIVKLAALRHYFT 1591
 QY 1588 NSWNIFDFVYVILSVGVVLSDIIOKTFPSPLFVYIRLARIQRIIRLIRGAKGIRTLIF 1647
 Db 1592 IGMNIFDFVYVILSVGVVLSDIIOKTFPSPLFVYIRLARIQRIIRLIRGAKGIRTLIF 1651
 QY 1648 ALMMSLPLFNLGILLFVMPFYSIFGMANFAYVYKMEAGIDMPNPOCFANMCLFOIT 1707
 Db 1652 ALMMSLPLFNLGILLFVMPFYSIFGMANFAYVYKMEAGIDMPNPOCFANMCLFOIT 1711
 QY 1708 TSAGMDGLSPILNTGPPYCDPTLPNSNGS--RGDCSPAVGILFTTYIISFLVYNNY 1766
 Db 1712 TSAGMDGLSPILNTGPPYCDPTLPNSNGS--RGDCSPAVGILFTTYIISFLVYNNY 1771
 QY 1767 IAIILNFSVATESBTEPLEDDFMFYEIWEKEDPEATQIIEYSVLSDFADALSEPLRI 1826
 Db 1772 IAIILNFSVATESBTEPLEDDFMFYEIWEKEDPEATQIIEYSVLSDFADALSEPLRI 1831
 QY 1827 AKPNQISILNMDLPMVSGDRHCHMDILFAFTRVYVSGEGEMALTIOMEERKMANPSKI 1886
 Db 1832 AKPNQISILNMDLPMVSGDRHCHMDILFAFTRVYVSGEGEMALTIOMEERKMANPSKI 1891
 QY 1887 SYEPIITTLRRKGEVSAMVIOARFRRHLQRLKHAFLFPOQAGSGISEBDAEREGT 1946
 Db 1892 SYEPIITTLRRKGEVSAMVIOARFRRHLQRLKHAFLFPOQAGSGISEBDAEREGT 1949
 QY 1947 IAYVNSNFSRPLGPPSSSSISSTSPPSYDSVTRATSDNIQVRGSDYSHSD 1999
 Db 1950 LIDKLNENST----PEKTDMPSTTSPPSYDSVTRATSDNIQVRGSDYSHSD 1999
 RESULT 26
 ADY27148
 ID ADY27148 standard; protein; 2005 AA.
 XX
 AC ADY27148;
 XX
 DT 05-MAY-2005 (first entry)
 XX
 DE Human SCN2A variant R223Q.
 XX
 KW SCN2A; anticonvulsant; muscular-Gen.; neuroprotective; antiarrhythmic;
 KW antismigraine; nootropic; antiparkinsonian; neuroleptic; tranquilizer;
 KW antidepressant; analgesic; nephrotoxic; antidiabetic; cyostatic;
 KW diagnostic; anxiety disorder; major depressive disorder; epilepsy;
 KW paralytic; hyperthermia; myasthenia gravis; heart arrhythmia; ataxia;
 KW migraine; Alzheimer disease; Parkinson disease; cystic fibrosis; pain;
 KW inflammation; polycystic kidney disease; phobia; schizophrenia;
 KW neuropathic pain; hyperglycemia; hyperinsulinemia; sodium channel;
 KW mutant.
 KW
 KW Homo sapiens.
 OS Synthetic.
 XX


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Db      1146 -----VDIGAPAEQGP-----VEPESELE-----PACFTED 1174
Qy      1171 CYRRCPCCAVDTTQAPGKWMMLRKTCHIVHSHFETITIMILLSCALAFEDITYLEE 1230
Db      1175 CYRKCCCOISIEEGKLMNMLRKCYKIVHNFETPIVMILLSCALAFEDITYEO 1234
Qy      1231 RRTIVLEAYADKMFYFVLEMLKMAVYGFKFTNMCWLDLIVSVLSVANTL 1290
Db      1235 RRTITMELYADKMFYFVLEMLKMAVYGFKFTNMCWLDLIVSVLSVANTL 1294
Qy      1291 GFAEMGPISKRLTLRALRLALSPREGKRVVNVNLVCAIPSIMVNLVCLIFMILFISM 1350
Db      1295 GYSEIGAIKSLRTLRRLRLRLSRREGKRVVNVNLVCAIPSIMVNLVCLIFMILFISM 1354
Qy      1351 GYNLFAGKFGRCINOTEGDPLNNTYTVNKSQCESL--NLTELGYMKVKNFQNVGAG 1407
Db      1355 GYNLFAGKFGRCINOTEGDPLNNTYTVNKSQCESL--NLTELGYMKVKNFQNVGAG 1411
Qy      1408 YVALLQVATFKGMMIMYAAVDSRGVEBPQWENLYMYTYFVIFIRGSEFTNLFTGV 1467
Db      1412 YVSLLOVATFKGMMIMYAAVDSRGVEBPQWENLYMYTYFVIFIRGSEFTNLFTGV 1471
Qy      1468 IIDNFNOOKKKLGGODIFMTBEOKKYNNAMKKLGSKKPKPIPRPLNKYQGFIFDIYKQ 1527
Db      1472 IIDNFNOOKKKLGGODIFMTBEOKKYNNAMKKLGSKKPKPIPRPLNKYQGFIFDIYKQ 1531
Qy      1528 AFDVTIMFLICLNMVYTMVETDQSEKINILAKINLFLVALFTGECIVKALBRHYET 1587
Db      1532 VEDISIMILICLNMVYTMVETDQSEKINILAKINLFLVALFTGECIVKALBRHYET 1591
Qy      1588 NSMNIFFDPRVYVLSVIGVYLSDIIOKYPSSFTLFRVIRARIGRIIRLRKAKGIRTLIF 1647
Db      1592 IGMNIFDFVYVLSVIGVYLSDIIOKYPSSFTLFRVIRARIGRIIRLRKAKGIRTLIF 1651
Qy      1648 ALMMSLPALENIGLLEFLVMPFYISFGMANFAYVYKAGIDIMFNFQFPAFNSMLCLFOIT 1707
Db      1652 ALMMSLPALENIGLLEFLVMPFYISFGMANFAYVYKAGIDIMFNFQFPAFNSMLCLFOIT 1711
Qy      1708 TSAGMDGLSPILNTGPPYCDPTLPNSNGS--RGDCGSPAVGILFTTYIIISFLIVNMY 1766
Db      1712 TSAGMDGLSPILNTGPPYCDPTLPNSNGS--RGDCGSPAVGILFTTYIIISFLIVNMY 1771
Qy      1767 IAILIENSVAATESETEPLSEDDFPMFYIRKPPBPATQFIRYSVLSFPAALSEPLRI 1826
Db      1772 IAILIENSVAATESETEPLSEDDFPMFYIRKPPBPATQFIRYSVLSFPAALSEPLRI 1831
Qy      1827 AKPNQISILNMDLPWVSGDRHICMDILFAFTKRVYVGESEGMALKIOMEKEMANPSKI 1886
Db      1832 AKPNQISILNMDLPWVSGDRHICMDILFAFTKRVYVGESEGMALKIOMEKEMANPSKI 1891
Qy      1887 SYEPIITTLRRKHEVSAMVIOARFRHLLQSLKHAFLFRQOAGSGISEDAEREGTL 1946
Db      1892 SYEPIITTLRRKHEVSAMVIOARFRHLLQSLKHAFLFRQOAGSGISEDAEREGTL 1949
Qy      1947 IAYVMSNPSPRLGPPSSSSISSTSPPSYDSVTRATSDNLCVRSQSDVSHSD 1999
Db      1950 LIDKLNENST---PEKIDMTPTSTSPSPSYDSVTRATSDNLCVRSQSDVSHSD 1995

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KW      diagnostic; anxiety disorder; major depressive disorder; epilepsy;
KW      paralysis; hyperthermia; amyotonia gravis; heart arrhythmia; ataxia;
KW      migraine; Alzheimer's disease; Parkinson's disease; cystic fibrosis; pain;
KW      inflammation; polycystic kidney disease; phobia; schizophrenia;
KW      neuropathic pain; hyperglycemia; hyperinsulinemia; sodium channel;
KW      mutin.
XX      Homo sapiens.
OS      Synthetic.
XX      Key
FH      Location/Qualifiers
FT      MISC-difference 1003
FT      /label= L1003I
FT      /note= "wild-type Leu substituted with Ile"
XX      WO2005014863-A1.
XX      17-FEB-2005.
XX      06-AUG-2004; 2004WO-AU001051.
XX      07-AUG-2003; 2003AU-00904154.
XX      (BION-) BIONOMICS LTD.
XX      Mulley JC, Harkin LA, Dibbens LM, Phillips HA, Heron SE;
XX      Berkovic SF, Scheffer IE, Davy A;
XX      WPI, 2005-195767/20.
XX      N-PSDB, ADY27077.
PT      Identifying subject predisposed to disorder associated with ion channel
PT      dysfunction, involves determining presence of specific mutation event in
PT      genes encoding ion channel subunits.
XX      Claim 20; SEQ ID NO 85; 347bp; English.
XX      PS
XX      This invention describes a novel method of identifying a subject
XX      predisposed to disorder associated with ion channel dysfunction
XX      comprising ascertaining whether at least one of the genes encoding ion
XX      channel subunits in the subject has undergone a mutation. The invention
XX      also describes 1) isolated nucleic acid molecules encoding an isolated
XX      polypeptide which is a mutant or variant ion channel subunit (including a
XX      mutant KCNQ2 subunit, where the mutation event has occurred in C terminal
XX      domain of the subunit and leads to disturbance in the calcium binding
XX      affinity of the subunit) and 2) an expression vector, cells, antibodies
XX      and a method for producing a non-human transgenic animal which are all
XX      used for screening of candidate pharmaceutical agents for diagnosing or
XX      treating epilepsy or a disorder associated with ion channel dysfunction.
XX      The mutation detected in the method disrupts the functioning of an
XX      assembled ion channel so as to produce an epilepsy phenotype in the
XX      subject or produces one or more disorders associated with ion channel
XX      dysfunction such as hyper- or hypo-kalemic periodic paralysis, myotonia,
XX      malignant hyperthermia, myasthenia, cardiac arrhythmias, episodic ataxia,
XX      migraine, Alzheimer's disease, Parkinson's disease, schizophrenia,
XX      anxiety, depression, phobic obsessive symptoms, neuropathic pain,
XX      inflammatory pain, chronic/acute pain, Bartter's syndrome, polycystic
XX      kidney disease, Dent's disease, hyperinsulinemic hypoglycemia of infancy,
XX      cystic fibrosis, congenital stationary night blindness and total color-
XX      blindness in the subject or to produce an epilepsy phenotype when
XX      expressed in combination with one or more additional mutations or
XX      variations in the ion channel subunit genes. The products of the
XX      invention have anticonvulsant, muscular-Gen., neuroprotective,
XX      antiarrhythmic, antimigraine, nociceptive, antiparkinsonian, neuroleptic,
XX      tranquilizer, antidepressant, analgesic, nephrotoxic, antidiabetic and
XX      cytosolic activity. This sequence represents a fragment of the human
XX      sodium ion channel subunit SCN2A encoded by exon 17 which contains the
XX      mutation L1003I.
XX      Sequence 2005 AA;
XX      Query Match 61.0%; Score 6393.5; DB 9; Length 2005;
XX      Best Local Similarity 62.1%; Pred. No. 0;

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Matches	1300;	Conservative	233;	Mismatches	359;	Indels	201;	Gaps	33;
Qy	5	LLPRTSSFRFRPTRESLAIEKMAEKQARSGTTLQESREGLPEBEAPRPOLDIQASKKL							64
Db	6	LVPSPDSFFRPTRESLAIEQRIAEERAKRP---							62
Qy	65	PDLYNPPQELIGLEBLEDPEYSQKTFIVLANKGKTFRPSATNALVVLSPFHIRAA							124
Db	63	PFIVGDIPEWVSVELEDLDPIYINKKTFIVLANKKALSRSFSAIPALVILTPFNIRKDA							122
Qy	125	VKILVHSLFNNLMIMCTIITNCVFMAQHPPEWTKVYEYTPALVYTESLVKILARFCLH							184
Db	123	IKILVHSLFNNLMIMCTIITNCVFMTMSNPDMTKVVEYTFPGIYTFESLIKILARFGCLF							182
Qy	185	APFLRDPWNMLDPSVIIMAYTTEFVDIGNVSALRTFVLRALKTISVISGLKTIYVALI							244
Db	183	DFTFLRDPWNMLDPIVITPAVYTEFVDIGNVSALRTFVLRALKTISVIGLKTIYVALI							242
Qy	245	QSVKTLADVMVLTVTCISVFALIGOLFMGNLRHCVR-----NFTALNGTNGSV							294
Db	243	QSVKTLSDVMILTVFCISVFALIGOLFMGNLRNKCLQWPPDNSSFELINTSF--FNNSL							300
Qy	295	EADGLVME-----SLDIYLSDBENYVLKNGTSDVLGNSSDAGTCCEGYRCLAKGNP							348
Db	301	DONGTTPKRTYSIFPMWDEYIEDKSHFYFLEGONDALLCGNSSDAQCEBGIYCVAKGNP							360
Qy	349	DHGYSFDSFAMAFIALFLMTQDCWERYOQTLSAGKIYIFPMLVIPLGSPYLVNLI							408
Db	361	NYGYSFDTFSWAFSLFLMTQDPWENLYQTLRLAGKIYIFPVLVIPLGSPYLVNLI							420
Qy	409	LAVNMAVYEQNOATIAETEEKERPOEAMEMLKREHEALTIR-----GV							453
Db	421	LAVNMAVYEQNOATLEAEQKAEFQQLBOLKQOEBQAAAAAASERDFSGAGTI							480
Qy	454	DTVSSSLEMSPLAVVNSHE---RSKRKRKMSGTEBCEGDRLPKSDSEDGPR-----							504
Db	461	GVFSSSSVASKLSKSEKELKNRKKKKKQKQSGEER--KNDRYLKSSESDSIRRKGRF							539
Qy	505	-----AMNHLISLTRGLSRTSMKPRSSGSIPTFRRR--DIGSEADPADDENS							549
Db	540	SLBGRSLTYEKRFSSPHQSLISIRGSLFSPRNSRALSFSFRGRANDIGSEMDPADDEHS							599
Qy	550	TAGESESHRTSLVLP--WPLRTSAQGGPSPTSA--PGHALHKKNGSTYDCNGVSLGA							606
Db	600	TFEDDSDRSDSLFVPHRGERHSNVSAQSRASRYLPIIPMNGKHSAYDCNGVSLVG-							658
Qy	607	GDPEATSPGSHLLRPVMLEHPDITTPSEEGPGQWLTQAPCVGFESEPGARQALSAV							666
Db	653	GPSTILTSAGQL-----PBGTTTERPI--RKRSSSYHVSMDLLEDPTSRQRAAGTA							708
Qy	667	SVLTSALEBELLESRRHKCPRCWNRLAQRYLIWECCLPMSIKQGVKLVVMDPFTDLITIMC							726
Db	709	SLITVMEELLESRRQCPRCWYKFAMCLIMDCPKWLKVKGLVNLVMDPFVDLAIATIC							768
Qy	721	LYLANTLPMALERTYNTSEFEEMLOQNGLVFTGIFPAEMTFKILADPPYTFQCGNNIPDS							786
Db	769	LYLANTLPMAMEHYPMTEOPSSVLSVGNLVFTGIFPAEMFLKIADPPYTFQCGNNIPDS							828
Qy	787	IYVILSLMEIGLSRMSNLSVLSRFLRLRYFKLAKSPPTLNTLTIKIGNSVGLAGNLTLYL							846
Db	823	FVLSLSLMEIGLANVBSLSVLSRFLRLRYFKLAKSPPTLNTLTIKIGNSVGLAGNLTLYL							888
Qy	847	AIIVFIFAVVGNQLFGKNYSE--LRDSDGLLPRMHMDPFAFLIIFRILCGEWIETMW							904
Db	889	AIIVFIFAVVGNQLFGKSYKECVCKISNDCELPRMHMDPFHSFLVFRVLGCEWIETMW							948
Qy	905	DCMEVSGGSLCLVPLLVNVTGNLVYLANFLALILSSFSADNLUTAPDEREMANNQIALA							964
Db	949	DCMEVAGGTMCTLVFMMVNVIGNLVYLANFLALILSSFSDDNLAAADDNENANNQIALAG							1008
Qy	965	RIORGLRFYKRTTWDCCGLLRQRPQKPAAL-----AAQGLPSCATPFYSPPPETE							1017
Db	1009	RMQGKIDFVKRIREF---IQKAFVAKQKALDEIKLELDLANKKQSCIS-----							1054
Qy	1018	KVPEPRKRETFEES-----QPGQGP-----GPPE--PVCYPIA							1050
Db	1055	-----NHTTLEIGKDLNLYLDNGITTSIGSSVEKIVVDESDYMSFINNPSLTVYPIA							1108
Qy	1051	VAESDITDQEREDENSLGTERESSEKQESQPVSGGPEAPPPDSRTWSQVSATASSEASAS							1110
Db	1109	VGESDPE-----NLTKEFSSSEDM-----ESKEKLNATSSSEGST---							1145
Qy	1111	QADWRQOKAEPPQAPCGGEFPEDSCSGSTADMTNTLLEQLPDLQDVKDPEDCCTEG							1170
Db	1146	-----VDIGAPAGEQPE-----VEPESIE-----PEACTED							1174
Qy	1171	CVRRPCCAVDITQAPKVMWRRLKTCYHIVESHMFETFIIFMLISSGALAFEDILYEE							1230
Db	1175	CVRRPKCCQSIIEGKGLMNNLKKTCYKVENHMFETFIYFMLISSGALAFEDILYEQ							1234
Qy	1231	RKTIKVLLEYADKMFYFVLEMLLKNVAVGFKKYFTNACWDLPLIVDSVLSLVANTL							1290
Db	1235	RKTIKMLEYADKVFYIFILEMLLKNVAVGFQYFTNACWDLPLIVDSVLSVLTANAL							1294
Qy	1291	GFAEMGPISIRTLRALPLRLALSFFGMRVYNVALYGAIPSIINNVLLVCLIFMLIFSIM							1350
Db	1295	GYSLEGAIKSIRTLRALPLRLALSFFGMRVYNVALYGAIPSIINNVLLVCLIFMLIFSIM							1354
Qy	1351	GVNLPAKGFRCINOTEGDLPIANTYINNTSOGESL---NLTEGLYMTKYVNFNDYGAG							1407
Db	1355	GVNLPAGFHYCINVTYTGEM--FDVSVVANNSECKALIESNOTAR--MKVAVNNDVGLG							1411
Qy	1408	YLALLQVATFGKMDIMYAAVDSRGYEBQPOMEYNLYIYFVIFIIIGSFPTLNLFIGV							1467
Db	1412	YLSLLQVATFGKMDIMYAAVDSRNVELQPCYEDNLVWLYIFVIFIIIGSFPTLNLFIGV							1471
Qy	1468	IIDPNQOKKLLGGODIFMTBEQKYYNANKLGSKKPQKPIRPLPKYCGFIDITYKQ							1527
Db	1472	IIDPNQOKKFFGGODIFMTBEQKYYNANKLGSKKPQKPIRPPAKPQGMVDFPYKQ							1531
Qy	1528	AFQDTIMEGLGNVNTVMVETDQSPKINILAKINILFVAFPGECIVKLAALRHYFT							1587
Db	1532	VFDISIMILICLNVTVMVETDQSOEWNTLNYINILFYLFGCEVYLKSLRIYFT							1591
Qy	1588	NSMNIIDFVVVILSIGVTSLDIIQKFFSPTLFRVIRLARIGRLIRLIRGAKGIRTLF							1647
Db	1592	IGNMIFDVVVILSIGVTFALIELEKVFVSPTLFRVIRLARIGRLIRLIRGAKGIRTLF							1651
Qy	1648	ALMMSLPALENIGLLFLVMTYISIFGMANPAYKMEAGIDMRFQTFANSMLCLPQIT							1707
Db	1652	ALMMSLPALENIGLLFLVMTYIAPGMSNPAVYKREVGIDDMENFETFGNSMTCLPQIT							1711
Qy	1708	TSAGMDGLSPIINTGPPYCDPLPNSNGS--RGDCGSPAVALIFFTYIIISPLIVVMY							1766
Db	1712	TSAGMDGLLAPILNSGPPCDPDKHGSSVKGCGNSVGIFFVSVIIISFLVILVMY							1771
Qy	1767	IATILENSVATESETEPLESDDDFMFEIWEKEDEPEATQPIEXSVLSDPADALSEPURI							1826
Db	1772	IATILENSVATESETEPLESDDDFEMFEWEKEDEPDATQPIEFKXUSDPAADALPELI							1831
Qy	1827	AKNQIISLINDLPMVSGDRICHMDILFAFTKRYLVSGESGEMDALKIOMEKFPMAAPSKI							1886
Db	1832	AKPNKVOQIAMDLPVNSGDRICHMDILFAFTKRYLVSGESGEMDALRIOMEERFMAAPSKV							1891
Qy	1887	SYEPITTLTKRKHEVSAWVQRAFRRLTORSLKHSFLRQQAQSGLSREDAAPERGL							1946
Db	1892	SYEPITTLTKRKQEVSAWVQRAFRRLTKQKAKVSSYIKKQKQKEC--DGTPIKEDT							1949
Qy	1947	IAYVSENFSPRLGPPSSSISSTSPSYSDVTRATSDNLQVNSDYSDSHSD							1999
Db	1950	LIDKLNENST-----PEKIDMTPTTSSPSSYSYVTKPEKEKE---KDKSEKD							1995
Qy	RESULT 28	ABB83627							
Db	ID	ABB83627							
		standard; protein; 2005 AA.							

XX AC AB883627;
 XX DT 10-OCT-2002 (first entry)
 XX DE Human GEFs+ protein with SCN2A mutation.
 XX KM Human; GEFs+, SCN2A, mutant; mletin;
 XX KM generalized epilepsy with febrile seizure plus.
 XX OS Homo sapiens.
 XX PN JP2002136289-A.
 XX PD 14-MAY-2002.
 XX PF 01-NOV-2000; 2000JP-00334969.
 XX PR 01-NOV-2000; 2000JP-00334969.
 XX PA (KAGA-) KAGAKU GIJUTSU SHINKO JIGYODAN.
 XX PA (RIKA) RIKAGAKU KENKYUSHO.
 XX DR WPI; 2002-552308/59.
 XX DR N-PSDB; ABQ79201.
 PT A human polynucleotide which is complementary to an mRNA transcribed from
 PT a generalized epilepsy with febrile seizure plus (GEFS+)-related gene
 PT useful for diagnosing GEFS+.
 XX PS Claim 10; Page 29-34; 37pp; Japanese.
 CC This invention relates to a human polynucleotide which is complementary
 CC to an mRNA transcribed from a "generalized epilepsy with febrile seizure
 CC plus" (GEFS+)-related gene. The gene is useful for diagnosing GEFS+. The
 CC present sequence represents the human GEFS+ protein sequence with SCN2A
 CC mutation
 XX SQ Sequence 2005 AA;
 Query Match 60.9%; Score 6392.5; DB 5; Length 2005;
 Best Local Similarity 62.1%; Pred. No. 0;
 Matches 1300; Conservative 232; Mismatches 360; Indels 201; Gaps 33;

421 LAVAMAAVEONQATLEAEAEQEAFFQMLDQLKKQGEAEQAQAAAAAASERDFSGAGSI 480
 454 DTSSRSLSLEMSPLAVNSHE--RRSKRKRMSSGTEBCGEDRLPKSDSESGPR----- 504
 481 GVFSSESSVSAKLSSEKELKNRRKKKKQKQEGSEEB-KDVRVAKSESEDSIRKGFRRF 539
 505 -----AMNHLSLRGLSRTSMKRRSSSGSI.FTPRR--DIGSEADPFADDESS 549
 540 SLEGSRLTYEKRFSPPHOSLSIRGSLFSPRRNSASLSPFRGAKDIDGSENDPFADDESS 599
 550 TAGESHSRTSLVY--WPLRRTSAQGPSPTSA-PGHALHGKKNSTVDCGVVSLIGA 606
 600 TFEEDNSRDLDFVHRHGERRHNSVQASASRLPLIPNNGKMSHSAVDCNGVSLVG- 658
 607 GDPKATSPGSHLRVPMLEHPPDTTSPSEEGPGQMLTSQAPCVDFEPEGARQALSAV 666
 659 GPSTLTSAGQL-----PEGTTETEI--RKRSSSYHSMDLBEDPSTRQRAMSIA 708
 667 SVLTSALBELFEESRHKCPKCNRLAQRLLIWECCPLMMSIQGVLYVMMDPDTLTIIMC 726
 709 SILNTMELEESRQKCPKCNRLAQRLLIWECCPLMMSIQGVLYVMMDPDTLTIIMC 768
 727 IVLNTLFMALRHYNMTSEFEEMLOVGNLVFTGIFTAEWTFKIALDPYYPQOGNNIPDS 786
 769 IVLNTLFMAHEHYPTTBQFSSVLSVGNLVFTGIFTAEWTFKIALDPYYPQOGNNIPDS 828
 787 IIVILSMEIGLSRMSNLVLSRFLRLRVFKLANSWPTLNTLKIIGNSVGLAGNLTVL 846
 829 FIVSLTMEIGLAVNEGSLVLSRFLRLRVFKLANSWPTLNTLKIIGNSVGLAGNLTVL 888
 847 AIIYPIPAVVGMOIFGKNYSF--LRDSGSLPRHMDPFHALPIIRIIGENIETMW 904
 889 AIIYPIPAVVGMOIFGKNYSF--LRDSGSLPRHMDPFHALPIIRIIGENIETMW 948
 905 DCMESVGSGLCLVFLVLMVIGNLVNLFLALILSSPADNLAPDRENNIQLALA 964
 949DCMEVAGQMLCLYFPMVAVIGNLVNLFLALILSSPADNLAPDRENNIQLALA 1008
 965 RIORGARFVAKRTTWDFCCGLLRORPQKPAAL-----AAGQGLPSCIATPYSPPPETE 1017
 1009 RMQGIDPVRKRIKF--IQKAFVRQKALDEIKPLEDINNKKDSCIS----- 1054
 1018 KVPPIPKRTREBE-----OPGQSTP-----GDPE-PYCVPIA 1050
 1055 -----NHTTBIGDNLVLYLDGNGTTSIGISSVEKYVDESDYMSFINNSLTVTVPIA 1108
 1051 VAESPTDQGEDEDESSSGTESESSKOSQAPVSGGEAPDPDSRTWSQVATSSSEAEASAS 1110
 1109 VGESDPE-----NLTBTEFSSBSDE-----ESKEKLNATSSBSGT--- 1145
 1111 QADRWQKAEPOAPGCGETPEDESCSEGSTADMTNTELBQIDPDGQDVDPEDCFTEG 1170
 1146 -----VDIGAPABEGQPE-----VEPESSLE-----PEACFIED 1174
 1171 CVRCPCCAVDTTQAPGKVMRLAKTYHYHESWFEFFIIFMILSSGALAFEDITYEB 1230
 1175 CVRKFCCQGISIEBGKGLMMNLKRTCYIYEHWFEEFFIIFMILSSGALAFEDITYEB 1234
 1211 RKTIVLEADKMTYFVYVEMLLKNVAYGFKKYFTNACWMLDPLIYDVSIVSLVANTL 1290
 1211 RKTIVLEADKMTYFVYVEMLLKNVAYGFKKYFTNACWMLDPLIYDVSIVSLVANTL 1290
 1235 RKTIVLEADKMTYFVYVEMLLKNVAYGFKKYFTNACWMLDPLIYDVSIVSLVANTL 1294
 1291 GFAEMGPIKSLRTLRALRPLALSRFBGMRVVAVNALVGAIPSSIMNVLLVCLIFMLIFSIM 1350
 1295 GYSELGAIKSLRTLRALRPLALSRFBGMRVVAVNALVGAIPSSIMNVLLVCLIFMLIFSIM 1354
 1351 GVNIPAGKFGKCIQTEGDLPLANTTYNNKQCSL--NLTGELYTKVAVNPDNVAG 1407
 1355 GVNIPAGKFGKCIQTEGDLPLANTTYNNKQCSL--NLTGELYTKVAVNPDNVAG 1411
 1408 YLALLQVATPKGMDIMVAAVDSRGYEQPQWENLVVYIYFVIFIGSFPTNLFLFQV 1467

Db 1412 YLSLQVATFKGMDIMTAANDSRNVEIQPKEDNLVWLYFVFIIFGSEFTLNLPIGV 1471
QY 1468 IIDNENQOKKKLGGODIEMTEBOKKYANMKKLGSKQKQPIPRELNTKYGFIEDIVTKQ 1527
Db 1472 IIDNENQOKKKRGGODIEMTEBOKKYANMKKLGSKQKQPIPRELNTKYGFIEDIVTKQ 1531
QY 1528 AFDVTIMELICLANTMTMVEETDDQSEKINILAKINLIFVALFPGECVXLALRHYET 1587
Db 1532 VFDISIMTILICLANTMTMVEETDDQSEKINILAKINLIFVALFPGECVXLALRHYET 1591
QY 1588 NSMNFDFVWVLTIVGTVLSIDIQYFESPTLFRVILARIGRLIRGAKGIRTLIF 1647
Db 1592 IGMNIFDFVWVLTIVGTVLSIDIQYFESPTLFRVILARIGRLIRGAKGIRTLIF 1651
QY 1648 ALMMSLPALFNIGLLFLVMPYISIFGMANFAYVMEAGIDMNFQTFPANSMLCLPQIT 1707
Db 1652 ALMMSLPALFNIGLLFLVMPYISIFGMANFAYVMEAGIDMNFQTFPANSMLCLPQIT 1711
QY 1708 TSAGMDGLSLPLNMPYCDPTLNSNGS-RGDCGSPAVGILEFTTYIIISFLVWNY 1766
Db 1712 TSAGMDGLSLPLNMPYCDPTLNSNGS-RGDCGSPAVGILEFTTYIIISFLVWNY 1771
QY 1767 IAIIEENFSVATEESTEPLEDDPMFYIEMKEFDEATQFIYESVLSDPADLSEPLRI 1826
Db 1772 IAVIIEENFSVATEESTEPLEDDPMFYIEMKEFDEATQFIYESVLSDPADLSEPLRI 1831
QY 1827 AKPNQISILIMDLPMVSGDRHICMDILFAFTRKVLGEGSEMALKIOMEKFMANPSKI 1886
Db 1832 AKPNQISILIMDLPMVSGDRHICMDILFAFTRKVLGEGSEMALKIOMEKFMANPSKI 1891
QY 1887 STEPIITTLARKHEEVSANVIGAPRRHILQSLKHAFLFRQAGSGISEEDAPERGL 1946
Db 1892 STEPIITTLARKHEEVSANVIGAPRRHILQSLKHAFLFRQAGSGISEEDAPERGL 1949
QY 1947 IAYVSENFSPRLGPPSSSISSTSPPSYDVTATSDNLQVRGSDVSHSD 1999
Db 1950 IAYVSENFSPRLGPPSSSISSTSPPSYDVTATSDNLQVRGSDVSHSD 1995

RESULT 29
ADB78604
ID ADB78604 standard; protein; 2005 AA.
XX ADB78604;
AC ADB78604;
DT 04-DEC-2003 (first entry)
XX
DE Human sodium channel subunit mutant SEQ ID NO:148.
XX
KW mutectin; mutant; ion channel; ion channel subunit; ICS; nootropic;
KW neuroprotective; inotropic; antipruritic; antiarthritic; antimalarial;
KW antidepressant; antiparkinsonian; neuroleptic; tranquilizer; analgesic;
KW nephrotoxic; antidiabetic; ophthalmological; epilepsy;
KW ion channel dysfunction; human.
XX
OS Synthetic.
OS Homo sapiens.
XX
PN WO2003008574-A1.
XX
PD 30-JAN-2003.
XX
PF 08-JUL-2002; 2002MO-AU000910.
XX
PR 18-JUL-2001; 2001AU-00006452.
PR 05-MAR-2002; 2002AU-00000910.
PR 13-MAY-2002; 2002AU-00002292.
XX
PA (BION-) BIONOMICS LTD.
PA (WALL/) WALLACE R W.
XX
PI Mollay JC, Harkin LA, Dibbens LM, Phillips HA, Heron SE,
Berkovic SF, Scheffer IE;

XX
DR MPI: 2003-239332/23.
DR N-PSDB; ADB78643.
XX
PT Identifying predisposition to an ion channel dysfunction, such as
PT periodic paralysis, cardiac arrhythmias, migraine, Alzheimer's disease,
PT schizophrenia, anxiety and depression, by detecting encoding-gene
PT mutation events.
XX
PS Claim 13; SEQ ID NO 148; 106pp; English.

The invention relates to a novel method for identifying a subject
CC predisposed to a disorder associated with ion channel dysfunction. The
CC method comprises ascertaining if at least one of the genes encoding ion
CC channel subunits (ICS) has undergone a mutation event so that a cDNA
CC derived from the subject has any of 134 nucleotide sequences. The method
CC of the invention has nootropic, neuroprotective, inotropic, antipruritic,
CC antiarthritic, antiparkinsonian, neuroleptic, tranquilizer, analgesic, and
CC neuroleptic, tranquilizer, analgesic, neuroprotective, antidiabetic, and
CC ophthalmological activity. A polynucleotide of the invention acts as an
CC ion channel agonist, or ion channel antagonist. The methods, isolated
CC nucleic acids, polypeptides, antibodies, selectively modified non-human
CC modulator of an ion channel, cells and genetically modified non-human
CC animal, are useful for the diagnosis and treatment of epilepsy and/or a
CC disorder associated with ion channel dysfunction, such as hyper- or hypo-
CC kalemic periodic paralysis, myotonias, malignant hyperthermia,
CC myasthenia, cardiac arrhythmias, episodic ataxia, migraine, Alzheimer's
CC disease, Parkinson's disease, schizophrenia, hyperkplexia, anxiety,
CC depression, phobic obsessive symptoms, neuropathic pain, inflammatory
CC pain, chronic/acute pain, Bartter's syndrome, polycystic kidney disease,
CC Dent's disease, hyperinsulinemic hypoglycaemia of infancy, cystic
CC fibrosis, congenital stationary night blindness and total colour
CC blindness. The present sequence represents a mutant protein of the
CC invention. The sequence data for this patent is not represented in the
CC printed specification, but was obtained in electronic format directly
CC from WIPO at ftp.wipo.int/pub/published_pat_sequences.
XX
SQ Sequence 2005 AA;

Query Match 60.9%; Score 6392.5; DB 7; Length 2005;
Best Local Similarity 62.1%; Pred. No. 0;
Matches 1300; Conservative 233; Mismatches 359; Indels 201; Gaps 33;
QY 5 LIPRTSSFRFRFTESLAIIEKMAEKOQARSTTLQESREGLPPEEARPOLDQASKTL 64
Db 6 LVPEGPDSFRFTESLAIIEKMAEKOQARSTTLQESREGLPPEEARPOLDQASKTL 62
QY 65 PDLGNPPOELIGLEDDLPFYSTQKTFYVLANGKTIIFRSATNALVLSPPHPIRRA 124
Db 63 PFYGDIPPEWVSPLLEDLDPEYINKKTFYVLANGKTIIFRSATPALYILTFNPIRRLA 122
QY 125 VKIIVHSLFNNLIMCTITLNCVPMAGQHPDPPWTKVETTFATYTFESLVYLARGFLH 184
Db 123 IKILVHSLFNNLIMCTITLNCVPMAGQHPDPPWTKVETTFATYTFESLVYLARGFLH 182
QY 185 AFTFLADPMNLDSFVIMATYTFEVDLGNVSALRTERFVALKTIISVIGLKTIVGALI 244
Db 183 DFTFLADPMNLDSFVIMATYTFEVDLGNVSALRTERFVALKTIISVIGLKTIVGALI 242
QY 245 QSVKGLADVMVLYVFCISVFPALIGLQFMGNLRIHKCYR-----NFTALNGTNGSV 294
Db 243 QSVKGLADVMVLYVFCISVFPALIGLQFMGNLRIHKCYR-----NFTALNGTNGSV 300
QY 295 EADGLVME-----SLDYLYSDPENYVLLKNGTSDVTLGNGSSDAGTCPEGYRLKAGBNP 348
Db 301 DONGTTFRRVYSIFPMWDEYIEDKSHFYFLBQNDALLGNSSDAGTCPEGYRLKAGBNP 360
QY 349 DHGYSFSPFAWAFALFRMLTQDCWERTYQOTLSAGKIYMFPMVLYFLGSFYLVNLI 408
Db 361 NYGYSFSPFAWAFALFRMLTQDCWERTYQOTLSAGKIYMFPMVLYFLGSFYLVNLI 420
QY 409 IAVVMAAYEONQATIAETEKEKRFQEMEMLKKEHEALTR-----GV 453


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QY 1468 IIDNPNQKKKGGGDI FMTTEQKRYNNAMKLGSKKKPKPIRPLINKYGFIFDIYTKQ 1527
DB 1472 IIDNPNQKKKGGGDI FMTTEQKRYNNAMKLGSKKKPKPIRPLINKYGFIFDIYTKQ 1531
QY 1528 APVVTMTFLCLAMVMTMTTDOQSPKINILAKINILPAITGGCTVLAALRHYYFT 1587
DB 1532 VPDISIMILCLAMVMTMTTDOQSPKINILAKINILPAITGGCTVLAALRHYYFT 1591
QY 1588 NSNNIDFVVVILSIYGVLSIIIOKYFSPPLFRVYRLARICRIILRLRGAGIRTLIF 1647
DB 1592 IGMNIDFVVVILSIYGVLSIIIOKYFSPPLFRVYRLARICRIILRLRGAGIRTLIF 1651
QY 1648 ALMMSLPALFNIGLILFLVWFYISFGMANFAVYKWEAGIDMFNFOTFANSMCLFQIT 1707
DB 1652 ALMMSLPALFNIGLILFLVWFYISFGMANFAVYKWEAGIDMFNFOTFANSMCLFQIT 1711
QY 1708 TSAQMDGLSPIINTGPPYCDPTLPNSNGS -RDCGSPAVGILFPTYYIIISFLIVNMY 1766
DB 1712 TSAQMDGLSPIINTGPPYCDPTLPNSNGS -RDCGSPAVGILFPTYYIIISFLIVNMY 1771
QY 1767 IATILENFSVATEESTEPLESEDDPMFYEIMWEKEDDEATQFIEYSVLSDPADLSRPLRI 1826
DB 1772 IATILENFSVATEESTEPLESEDDPMFYEIMWEKEDDEATQFIEYSVLSDPADLSRPLRI 1831
QY 1827 AKPNQIQLINMOLPMVSGRIHOMDILFAFTKRVLGESGEMDLAKIOMEKFFMAANPSKI 1886
DB 1832 AKPNQIQLINMOLPMVSGRIHOMDILFAFTKRVLGESGEMDLAKIOMEKFFMAANPSKI 1891
QY 1887 STEPIITTTLRKHEEVSAMVIOQAFRRHLIQRSLKIASPLFRQAGSGLESDAPEREGI 1946
DB 1892 STEPIITTTLRKHEEVSAMVIOQAFRRHLIQRSLKIASPLFRQAGSGLESDAPEREGI 1949
QY 1947 IAVWSENFSPRPSSSSISSTSPSPYDSTVTRATSNLQVRGSDYHSHPD 1999
DB 1950 IAVWSENFSPRPSSSSISSTSPSPYDSTVTRATSNLQVRGSDYHSHPD 1999

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RESULT 31
ADB78605
ID ADB78605 standard; protein; 2005 AA.

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AC ADB78605;
DT 04-DEC-2003 (first entry)
XX
DE Human sodium channel subunit mutant SEQ ID NO:149.
XX
KW mutain; ion channel; ion channel subunit; ICS; nootropic;
KW neuroprotective; inotropic; antipyretic; antiarrhythmic; antimigraine;
KW antidepressant; antiparkinsonian; neuroleptic; tranquilliser; analgesic;
KW neurotropic; antidiabetic; ophthalmological; epilepsy;
KW ion channel dysfunction; human.
XX
OS Synthetic.
OS Homo sapiens.
XX
PN WO2003008574-A1.
XX
PD 30-JAN-2003.
XX
PF 08-JUL-2002; 2002WO-AU000910.
XX
PR 18-JUL-2001; 2001AU-00006452.
PR 05-MAR-2002; 2002AU-00000910.
PR 13-MAY-2002; 2002AU-00002292.
XX
PA (BION-/) BIONOMICS LTD.
PA (MALL-) WALLACE R W.
XX
PI Mulley JC, Harkin LA, Dibbens LM, Phillips HA, Heron SE,
PI Berkovic SF, Scheffer IE;
XX
DR WPI; 2003-239332/23.

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DR N-PSDB; ADB78644.
XX
PT Identifying predisposition to an ion channel dysfunction, such as
PT periodic paralysis, cardiac arrhythmias, migraine, Alzheimer's disease,
PT schizophrenia, anxiety and depression, by detecting encoding-gene
XX mutation events.
PS Claim 13; SEQ ID NO 149; 1066p; English.
XX
CC The invention relates to a novel method for identifying a subject
CC predisposed to a disorder associated with ion channel dysfunction. The
CC method comprises ascertaining if at least one of the genes encoding ion
CC channel subunits (ICS) has undergone a mutation event so that a cDNA
CC derived from the subject has any of 134 nucleotide sequences. The method
CC of the invention has nootropic, neuroprotective, inotropic, antipyretic,
CC antiarrhythmic, antitachycardic, antidepressant, antiparkinsonian,
CC neuroleptic, tranquiliser, analgesic, nephroretic, antidiabetic, and
CC ophthalmological activity. A polynucleotide of the invention acts as an
CC ion channel agonist, or ion channel antagonist. The methods, isolated
CC nucleic acids, polypeptides, antibody, selective agonist, antagonist or
CC modulator of an ion channel, cells and genetically modified non-human
CC animal, are useful for the diagnosis and treatment of epilepsy and/or a
CC disorder associated with ion channel dysfunction, such as hyper- or hypo-
CC kalemic periodic paralysis, myotonia, malignant hyperthermia,
CC myasthenia, cardiac arrhythmias, episodic ataxia, migraine, Alzheimer's
CC disease, Parkinson's disease, schizophrenia, hyperplexia, anxiety,
CC depression, phobic obsessive symptoms, neuropathic pain, inflammatory
CC pain, chronic/acute pain, Bartter's syndrome, poly cystic kidney disease,
CC Dent's disease, hyperinsulinaemic hypoglycaemia of infancy, cystic
CC fibrosis, congenital stationary night blindness and total colour
CC blindness. The present sequence represents a mutant protein of the
CC invention. The sequence data for this patent is not represented in the
CC printed specification, but was obtained in electronic format directly
CC from WIPO at ftp.wipo.int/pub/published_pat_sequences.
XX
SQ Sequence 2005 AA;

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Query Match 60.9%; Score 6391.5; DB 7; Length 2005;
Beat Local Similarity 62.1%; Pred. No. 0;
Matches 1300; Conservative 232; Mismatches 360; Indels 201; Gaps 33;

QY 5 LIPRGTSFRFTRESLAIEKMAEKQARGSTTLQESREGHPREEAPRPQDLQASKTL 64
DB 6 LVPPGPDSPFRFTRESLAIEKMAEKQARGSTTLQESREGHPREEAPRPQDLQASKTL 62
QY 65 PDLYGNPQELIGLEPLDLDPPYSTQKTFIVLNKKTIFRSATNALVYLSFPHPIRRA 124
DB 63 PRYGDIPPEWVSVPLELDLDPYINKKTFIVLNKKAISRSFATPALVILTFPNPIRKA 122
QY 125 VKIIVHSLFNNMLIMCTIITLNCVFMQOHDPPTKRYVETFTAIYFESVYKILANGFCIL 184
DB 123 IKILVHSLFNNMLIMCTIITLNCVFMQSNPDDTKNVEYFGIYTFESLKIILANGFCIL 182
QY 185 AFTFLRDPWNNLDPFVIIIMAAVYTBVDLGNVSALRTFRVLRALKITISVIGKTIYVGLI 244
DB 183 DFTFLRDPWNNLDPFVIIIMAAVYTBVDLGNVSALRTFRVLRALKITISVIGKTIYVGLI 242
QY 245 QSVKGLADVMVLTVCISVPAIIGLQTFMGNLRHKVR-----NFTALANGTNGSV 294
DB 243 QSVKGLADVMVLTVCISVPAIIGLQTFMGNLRHKVR-----NFTALANGTNGSV 294
QY 295 EADGLVME-----SLDIYLDPENYLLKNGSDVLTLCGNSDDACTCEGRCCLAKGENP 348
DB 301 DNGGTFNRTVSIFFWDEYIEKSHFYFLLEGQNDLALCGNSDDACTCEGRCCLAKGENP 360
QY 349 DHGYSFDSFAWAFALFRMTQDCWRLYOOTLSAGKIYVIFMLYIFLSSFLVNLVI 408
DB 361 NNGYTSFDFSMARSLFRMTQDCWRLYOOTLSAGKIYVIFMLYIFLSSFLVNLVI 420
QY 409 IAVVMAVEBONQATIAETEKEKRFQEAEMLKKEHEALYIR-----GV 453
DB 421 IAVVMAVEBONQATIAETEKEKRFQEAEMLKKEHEALYIR-----GV 453

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QY 454 DTVSRSLSIEMSPPLAVNSHE--RRSKRRKMSGTEECGBDRLEPKSSENGPR----- 504
 DB 481 GVFSSESSVASKLSKSEKELKNRRKKKKOKEOGSEER-KODRVLKSESEOSIRKGRF 539
 QY 505 -----ANNHSLTRGLSRTSMKPRSSGSIPTPRR--DGSEADPADENS 549
 DB 540 SLEGRLLTYEKRFSPHOSLSIRGSLSPRRNSPALSFRGRKADIGSEIDPADDHS 599
 QY 550 TAGSESHRTSLIV--WPLRRTSAOQOPSGTSA-PGHALHGKNSYVDCGVVSLGA 606
 DB 600 TFEODSDRDLFLVHRBGERHSNVSOASRASRLPLPMNGKMSAIVDCGVVSLVG- 658
 QY 607 GDPBATSFGSHLRVLMLEHPPDTTPPSSEPGPQMLTSQAPCVUGFEFGARQALSAV 666
 DB 659 GPSTLTSAGQL-----PEGTTTEI--RKRRSSSYHVSMDLIDPTSRQAMSAIA 708
 QY 667 SVLTSALEBELSESRHKCPPCNRRLAQRVLIWECPLMNSIKQGVKLVVMDPTDLEITMC 726
 DB 709 SLTNTMBELSESRKQCPPCMYKFAVMCLIMDCCPMLKVKLVLVMDPVDLALITC 768
 QY 727 IVLNTLFMALEHYNTSEFEEMIQVGNLVFTGIFTAEMTFKIIALDPYIYFOQWNIFDS 786
 DB 769 IVLNTLFAMMEHYPTTEQSSVLSVGNLVFTGIFTAEMFLKIIAMPYIYFOQWNIFDG 828
 QY 787 IIVILSLMELGSRMSNLSVLSFLRLRFKIAKSPITNTLITIGNSVGLAGMLTYL 846
 DB 829 FIVSLIMELGLANVGLSVLSFLRLRFKIAKSPITNTLITIGNSVGLAGMLTYL 888
 QY 847 AIIIVFAVVGQOLFQKNSY--LRDSGGLPRHMDPFPAFLIIFILGEMILETMW 904
 DB 889 AIIIVFAVVGQOLFQKNSY--LRDSGGLPRHMDPFPAFLIIFILGEMILETMW 948
 QY 905 DCMENVSGOSLCILVFLVNVGNLVVNLFLALLSSFSADNLAPDEDEKNNIQLALA 964
 DB 949 DCMENVAGQIMCLTVFEMVWVIGNLVYVNLFLALLSSFSADNLAPDEDEKNNIQLAIVG 1008
 QY 965 RIQRGLRFYKRTTWDFCCGLLRORPOKPAAL-----AAQQLPBCIATPSPPEPTE 1017
 DB 1009 RMQKGIDFYKRXIREP---IQKAFVRKQALDEIKLELDLNNKQSCIS----- 1054
 QY 1018 KVPTRKETRPEGE-----OPQGT-----GDPE-PVCVPIA 1050
 DB 1055 -----NHTTIEIGKDLNTYKDGANGTISGIGSEVKEYVVDSESYMFINNPSLTYVPIA 1108
 QY 1051 VASDITDDEDEBENSAGTEBESSKQESQPVSGPAPDSRTSOGVASTASSEASAS 1110
 DB 1109 VGSDDP-----NLNTEEFSSSEDM-----ESKEKLNTATSSSEGST-- 1145
 QY 1111 QADMROQMAEPAPGCGETPBDSCSEGSTADMNTAELLBOJPDIGQVQDECFTEG 1170
 DB 1146 -----VDIGAPAEGEQPE-----VEBESLE-----PEACFTEG 1174
 QY 1171 CVARCPCAVDTTQAQKVMWMLRKTGYHIVESHMFETFIIMILSSGALAFEDIYLEE 1230
 DB 1175 CVARKECCQISIEGSKLMMNLRKTCYKVEHNMETPIVEMILSSGALAFEDIYIQ 1234
 QY 1231 RKTIKVLEBYADKFTYFVLEMLKMAVAGFKKYFTNAWCMLDPLIIVSVLSVANTL 1290
 DB 1235 RKTIKMLBYADKFTYFIFLEMLKMAVAGFOVFTNACWMDPLIIVSVLSVANTL 1294
 QY 1291 GFAEMGPISKLRTLRALRPLRALSREPGMAYVNVNVALVGAIPSIINVTLVCLIFMILPSIM 1350
 DB 1295 GYSELGAIKSLRTRALRPLRALSREPGMAYVNVNVALVGAIPSIINVTLVCLIFMILPSIM 1354
 QY 1351 GVNLPAGKFGRCINOTEGDLPLNYTIVNKSOCESL---NLGELVYTKVKNFVNDVAG 1407
 DB 1355 GVNLPAGKFGYHCINYTTEGM-FDVSVNVNYSSEKALIESQOTAR--WKVKNFVNDVAG 1411
 QY 1408 YLALLQVATPKGMNDIMYAAVDSRGVEBQPMQEMNLYMYIYFIIIFGSEFTLNFITGV 1467
 DB 1412 YLALLQVATPKGMNDIMYAAVDSRGVVELQPKYEDNLMYLYFYIIFIGSEFTLNFITGV 1471
 QY 1468 IINFNQKKKGLGGQDIFMTBQKTYNNAKKLGSKKQKPIRPLANKYGFIDIVTKQ 1527

DB 1472 IINDFNQKKKGGQDIFMTBQKTYNNAKKLGSKKQKPIRPLANKYGFVDFVTKQ 1531
 QY 1528 AFDVTIMFLICLNNVTMMVETDDOSPEKINIILAKINILFPAIFGCEIVLAAIRHYFT 1587
 DB 1532 VFDISIMILICLNNVTMMVETDDOSQEMTILVWINIVFTLVFGCEIVLAKISIRHYFT 1591
 QY 1588 NSNNIPFVVVYILSVGVVLSDIQKFFSPFLFRVIRLARIGILRLIRGAKGIRTLF 1647
 DB 1592 IGMNIPFVVVYILSVGVVLSDIQKFFSPFLFRVIRLARIGILRLIRGAKGIRTLF 1651
 QY 1648 ALMMSLPALFNIGLLFLVMEIYISIFGMANFAYVWKEAGIDDMNFOTFANSMLCLFOIT 1707
 DB 1652 ALMMSLPALFNIGLLFLVMEIYIAIFGMSNFAVYKREVGIDDMNFOTFANSMLCLFOIT 1711
 QY 1708 TSAGMDGLSLPILNTGPPYCDPTLPNSNGS-RGCGSPAVGIIIFETYYIIISFLVNMV 1766
 DB 1712 TSAGMDGLSLPILNTGPPYCDPTLPNSNGSVKGGCGNPSVGIFFVSTIIISFLVNMV 1771
 QY 1767 IAILNENSVATERSTPELSDDFPMFEIWEKFPPEATOPFEYSVLSDFADALSEPLRI 1826
 DB 1772 IAILNENSVATERSTPELSDDFPMFEIWEKFPPEATOPFEYSVLSDFADALSEPLRI 1831
 QY 1827 AKPNQISLINDLPMVSGDRICHMDILPAFTKRVLGSEGENDALKIQMEKFMALNPSKI 1886
 DB 1832 AKPNQVOLIAMDLPVSGDRICHMDILPAFTKRVLGSEGENDALKIQMEKFMALNPSKV 1891
 QY 1887 SYEPIITTLRKKEHVSAMVQRAFRRLRLORSILKHSFLRQQAQSGLSREDAPERGL 1946
 DB 1892 SYEPIITTLRKKEHVSAMVQRAFRRLRLORSILKHSFLRQQAQSGLSREDAPERGL 1949
 QY 1947 IAYVSENFSPRLGPPSSSISSTSPSPSYSTRATSDNLQVSGDSYSHSD 1999
 DB 1950 LIDKLINENST---PEKIDMTPTTSPSPSYSTRATSDNLQVSGDSYSHSD 1995

RESULT 32

ADCA6947 standard; protein; 2005 AA.

ADCA6947;
 18-DEC-2003 (first entry)

Human SCN2A amino acid sequence #SEQ ID 3.

SCN2A; voltage-gated ion channel; human; neuroprotective; gene therapy;
 vaccine; Alzheimer's disease.

Homo sapiens.

WO2003060525-A1.

24-JUL-2003.

16-JAN-2003; 2003WO-EP000400.

17-JAN-2002; 2002EP-00001236.

17-JAN-2002; 2002US-0348674P.

(EVOT-) EVOTEC NEUROSCIENCES GMBH.

Hipfel R, Von Der Kammer H, Pohlner J;

WPI; 2003-598580/56.

N-PSDB; ADCA6947.

Diagnosing or prognosticating a neurodegenerative disease by detecting the level or activity of transcription or translation products of the gene coding for the voltage-gated ion channel SCN2A.

Disclosure; Fig 9; 67pp; English.

XX

Db	1832	AKPNKVOLIAMDLPWVSGDRHICLDILFAFTKRVLVGSGEMDALRIQMERFMAANSKY	1891
Qy	1887	SYEPITTLRRKHEEVSAMVIQAFRRHLQRLKHSFLFRQAGSLSEDAPEREG	1946
Db	1892	SYEPITTLTKRKQREVSAILIQRAYRRLKQKVKVSIYKKQKKEC--DGTPIKEDT	1949
Qy	1947	IAYVSNFSRPLGPPSSSISSISFPSPSYSVTRATSDNIQVGSYSHSED	1999
Db	1950	LIDKLINENST----PEKDTMPTSTTSPSYSDSVTKPEKEKPE---KKSKEXED	1995
RESULT 33			
ID	ADSS2265	standard; protein; 2005 AA.	
XX	ADSS2265		
AC	ADSS2265;		
DT	30-DEC-2004	(first entry)	
XX			
XX		Human sodium channel type II (SCN2A) protein SeqId60.	
DE			
XX			
KW		sodium channel type II; SCN2A; mutation; mis-sense; nonsense;	
KW		frame shift; splice site; obstinacy childhood epilepsy; mental illness;	
KW		human.	
XX			
OS		Homo sapiens.	
XX			
PN	JP2004275115-A.		
XX			
PD	07-OCT-2004.		
XX			
PF	18-MAR-2003; 2003JP-00072979.		
XX			
PR	18-MAR-2003; 2003JP-00072979.		
XX			
PA	(DOKU-) DOKURITSU GYOSEI HOJIN RIKAGAKU KENKYUSH.		
PA	(KOKU-) KOKURITSU RYOOTO SHIZUOKA SHINKAI IRYO.		
XX			
DR	WPI; 2004-712375/70.		
XX			
DR	N-PSDB; ADSS22606.		
XX			
PT	Novel isolated human sodium channel type II SCN2A gene, its cDNA or mRNA		
PT	having disease mutation e.g., mis-sense, nonsense, frame shift or		
PT	splicing site mutation, useful as primer or probe for detecting obstinacy		
PT	childhood epilepsy.		
XX			
PS	Disclosure; SEQ ID NO 60; 59pp; Japanese.		
XX			
XX			
CC	This invention relates to a novel isolated human sodium channel type II		
CC	(SCN2A) gene, its corresponding cDNA or mRNA, having a disease mutation		
CC	(mis-sense, nonsense, frame shift, splicing site mutation) in the DNA		
CC	which encodes the human SCN2A protein. The invention enables diagnosis of		
CC	obstinacy childhood epilepsy accompanying regression of serious mental		
CC	illness, by detection of a mutation in a gene, and thus contributes to		
CC	the development of treatment methods. The present sequence is that of the		
CC	protein encoded by the human sodium channel type II (SCN2A) gene of the		
CC	invention.		
SQ	Sequence 2005 AA:		
Query Match 60.9%; Score 6391.5; DB 8; Length 2005;			
Best Local Similarity 62.1%; Pzed. No. 0;			
Matches 1300; Conservative 232; Mismatches 360; Indels 201; Gaps 33			
Qy	5	LLRRGTSFRFRFRESIAATEKMAEQAGSTTIGSREGLPEFEAPRPQDLQASKYL	64
Db	6	LVPPGSPSFRFPRESIAALEQRRAEKAKRP---KQERQDDDEGPKPNSDLEAGKSL	62
Qy	65	PDLVGNPOELIGEDLDLDFEYSTQCTFTVLNKGKTIFFRSATNLVYLSPHPPIRRAA	124
Db	63	PRFYGDLPPEKMSVSLPDDLDPPYINKTKFTVLNKGKALISFSMTPLYLITPNNPIRKLA	122
Qy	125	VKLIVHSLFNMLINCTILTNCFVMAQHDPPPWTKIVYEYTFTAITYTESLVKIIARGFCLH	184

Db	123	IKLIVSLIFMMLIMCILLITNCVFMVMSNPBDTKVNEYFTGAIYFPESILIKLILANGFCLE	182
Qy	185	AFFTELDPMMWULDPESYIIMAYTTEFPDLDGNVSALRFRVLRALKTISVISGLTIVGALI	244
Db	183	DFFELRPMWMDPFTVITRAYTEFEFDLDGNVSALKRFRVLRALKTISVIRGLKTIWGLI	242
Qy	245	QSVKELADVMVLTVFCLSVPFALIGLOFMGNLRHKVR-----NFTLANGNSV	294
Db	243	QSVKELADVMVLTVFCLSVPFALIGLOFMGNLRHKVR-----NFTLANGNSV	300
Qy	295	EADGLWE-----SLDLYISDPENYLLKNGTISDVLICGNSSDAGCPGKYCLAGENP	348
Db	301	DGNGTTFNRTVSIJFNWDEYIEDSHFYLEGQNDALLCNSSDAGCPGKYCVAGRNP	360
Qy	349	DHGTSFDSFAMFLIFRLMTODCWBRLYQOULRSAGKIYIMFMLVYFLGSFYLWLI	408
Db	361	NYGTSTPDTSSWALSLFRMTODFBNLYQULTRAGKTYMFFVLVIFLGSFYLIWLI	420
Qy	409	LAVAMAYEONQATTIETEEKERKPEJAMWMLKKEHALTIR-----GV	453
Db	421	LAVAMAYEONQATTIETEEKERKPEJAMWMLKKEHALTIR-----GV	480
Qy	454	DTVSRSSLIENSPLAPVNSH-----RRSKRRKRMSSGTEBCEBDLPSPSDEDDR	504
Db	481	GVFSSSSSVASKLSSEKELKNRRKKKKOKESGEE-----KNDVLKSEBDSIRKKGFRP	539
Qy	505	-----AMNLSLIRGLSRTSMKPRSSRGSIFTFRRR-----DLGSEADPADENS	549
Db	540	SLEGSRLTYEKRFSSPHOSLISTIRGLSFRSRKRSKSLPFRGRADISENDFADENS	599
Qy	550	TAGESESHRTSILVP-----WPLKRTSAQOPSPGTS-----PGHALHCKKNSYDNCNGVSLGQ	606
Db	600	TFEENDSRDRLFPHRHGERRRHSNVQASRASRVLPILPMNGKMSAVIDCNGVSLVG-----	658
Qy	607	GDPEATSPGSHLPRVMLNHPDPTTTSSEBPGPOMLTQAPCVDFEERPGARORLASV	666
Db	659	GPSTLTSAGULL-----PEGITTEIETI-----KRRSSYHVSMDLIEDTSSQRMASIA	708
Qy	667	SVTSSALBELSESRHKPCPCWNRBLAQOYLIMECCEPLMMSIKQVKLVWMDPFTDLTTTC	726
Db	709	SILNTMTELESSEKOKPCPCMYKALMCLIMDCCKPKXKHLVNLVWMDPFDLAIITIC	768
Qy	727	IYANTLPMALBRYMNTSEFEEMLOVGNLVFTGIFTAEMTEKIIALDYYYYFOGQWNI	786
Db	769	IYANTLPMALBRYMNTSEFEEMLOVGNLVFTGIFTAEMTEKIIALDYYYYFOGQWNI	828
Qy	787	IIVILSIMEIGLSRMSNLSVLRFRLLRVFLKASWPTLNTLKIIGNSVGAIGNLTLVL	846
Db	829	FIVSLIMELGIANVEGILSVLRFRLLRVFLKASWPTLNTLKIIGNSVGAIGNLTLVL	888
Qy	847	AIIVITFVAVNGOLFGKNYS-----LRDSOGLPMHMDPFHAFLIFIRLICEGMIETW	904
Db	889	AIIVITFVAVNGOLFGKNYS-----LRDSOGLPMHMDPFHAFLIFIRLICEGMIETW	948
Qy	905	DCMEVSGOSLCLVFLVAVNGVNLVNLFLATLLISFSADNTLTAPEDEKMNLOLALA	964
Db	949	DCMEVAGQTMCLTYMNMVAVNGVNLVNLFLATLLISFSADNTLTAPEDEKMNLOLALA	1008
Qy	965	RIORGRLPVAKRTWDFCCGLLRQRPQAPAL-----AAQOLPSCIATPIYSPPEETE	1017
Db	1009	RMQKGIDFVKRIREF-----IQKAFRKOKALDEIKPLEDLNNKDOCSIS-----	1054
Qy	1018	KVPPIRKEKREFGE-----QPGQGP-----GDPE-----PVCVPIA	1050
Db	1055	-----NHTTIEIGLDNLYLKDNSTTSGTSSVEKYVVDSDYMSFLNPSLTVYPIA	1108
Qy	1051	VASDPTDDEBDEENSLGTEESRSKQESQPVSGGEAPPPDSRTWSQVSATASSAEASAS	1110
Db	1109	VGSBDE-----NLTREBSSSDME-----ESKXKLNATSSSEGST---	1145
Qy	1111	QADWROOWAEOAPQCGEFTPDCSCBSSTADMTNTAELQIPLQGDYVDQPEDCFTBEG	1170

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Db 1146 -----VDIGAPAGEOPE-----VEPESELE-----PEACPTED 1174
Qy 1171 CVBRCCCAVDITQAPCKWMMRLKTCYHIVESHWEFTIIFMILLSSGALAEEDYLEE 1230
Db 1175 CVRRPKCCQISIEGKLMWNLKTCYKIVENHWPEFTIVFILLSSGALAEEDYLEE 1234
Qy 1231 RKTIKVLLEADKMFYVFLVLEMLKVAAGFKKYPFNACMDLFLVDVSLVSVANTL 1290
Db 1235 RKIKMLLEADKMFYVFLVLEMLKVAAGFKKYPFNACMDLFLVDVSLVSVANTL 1294
Qy 1291 GFAEMGPIKSLRTLRALRPLRALSREFGMRVVNVALGALPSIMNVLLVCLIFMLIFSIM 1350
Db 1295 GYSELGAIKSLRTLRALRPLRALSREFGMRVVNVALGALPSIMNVLLVCLIFMLIFSIM 1354
Qy 1351 GNVLPFGKRCRCINQREBGLPLATYIVNKSOCESL---NLGELVWTKRKNPDPVAG 1407
Db 1355 GNVLPFGKRCRCINQREBGLPLATYIVNKSOCESL---NLGELVWTKRKNPDPVAG 1411
Qy 1408 YLALLOVATEFGKMDIMYAADSRGYEEOQPMENLYMYIYFIIFGSPFLNLPFGV 1467
Db 1412 YLSTLQVATFGKMDIMYAADSRGYEEOQPMENLYMYIYFIIFGSPFLNLPFGV 1471
Qy 1468 IIDNFQOQKKKLGQDI FMTBEOKKYNNAMKLGSKKPKQPIRPLNKYOGFIDIVTKQ 1527
Db 1472 IIDNFQOQKKKLGQDI FMTBEOKKYNNAMKLGSKKPKQPIRPLNKYOGFIDIVTKQ 1531
Qy 1528 APDVIMELLCLMMVTMMVETDQSPKINIILAKINLLFAITGECIVGLALRRHYPT 1587
Db 1532 VEDISIMILLCLMMVTMMVETDQSPKINIILAKINLLFAITGECIVGLALRRHYPT 1591
Qy 1588 NSNNIDPFVNVLSIYGVTVLSIIIOKFFSPTLFVRILARIRILKIGAKGITLLE 1647
Db 1592 IGNITDFVNVLSIYGVTVLSIIIOKFFSPTLFVRILARIRILKIGAKGITLLE 1651
Qy 1648 ALMMSLPALFNIGLLFLVWFYISIFGMANFAYVKKWAGIDMNFQTFANSMCLFOIT 1707
Db 1652 ALMMSLPALFNIGLLFLVWFYISIFGMANFAYVKKWAGIDMNFQTFANSMCLFOIT 1711
Qy 1708 TSGMGGLSPIANTGPPYCDPLPNSNGS-RGDCSPAVGLIFFTYIIISFLYVNMV 1766
Db 1712 TSGMGGLSPIANTGPPYCDPLPNSNGS-RGDCSPAVGLIFFTYIIISFLYVNMV 1771
Qy 1767 IATILENFVATEESTEPSEDEDFMFEIWEKEDPEAFOFIYSVLSPADLSPLRI 1826
Db 1772 IATILENFVATEESTEPSEDEDFMFEIWEKEDPEAFOFIYSVLSPADLSPLRI 1831
Qy 1827 AKNQSLINMDLPMVSGDRHICMDLFAFTKRVLGESGEMDLAKTOMEKFFMANPSKI 1886
Db 1832 AKNQSLINMDLPMVSGDRHICMDLFAFTKRVLGESGEMDLAKTOMEKFFMANPSKI 1891
Qy 1887 STEPIITTLRKHEEVSAMVIOAPRRHLQSLKIASFLFROAGSSGLSEEDAPREGL 1946
Db 1892 STEPIITTLRKHEEVSAMVIOAPRRHLQSLKIASFLFROAGSSGLSEEDAPREGL 1949
Qy 1947 IAYVSENSRPLGPPSSSISSTSPSYDSVTRADNLQVRGSDYSHSD 1999
Db 1950 IAYVSENSRPLGPPSSSISSTSPSYDSVTRADNLQVRGSDYSHSD 1995

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KW paralytic; hyperthermia; myasthenia gravis; heart arrhythmia; ataxia;
KW migrating; Alzheimers disease; Parkinsons disease; cystic fibrosis; pain;
KW inflammation; polycystic kidney disease; phobia; schizophrenia;
KW neuropathic pain; hyperglycemia; hyperinsulinemia; sodium channel;
KW mutein.
XX Homo sapiens.
OS Synthetic.
FH Key Location/Qualifiers
FT MISC-difference 1319
FT /label= R1319Q
FT /note="wild-type Arg substituted with Gln"
XX
XX WO2005014863-A1.
XX
XX 17-FEB-2005.
XX
XX 06-AUG-2004; 2004WO-AU001051.
XX
XX 07-AUG-2003; 2003AU-00904154.
XX
XX (BION-) BIONOMICS LTD.
XX
XX Muller JC, Harkin LA, Dibbens LM, Phillips HA, Heron SE;
XX Berkovic SF, Scheffer IE, Davy A;
XX WPI, 2005-195767/20.
XX N-PSDB; ADY27079.
XX
XX PT Identifying subject predisposed to disorder associated with ion channel
XX PT dysfunction, involves determining presence of specific mutation event in
XX PT genes encoding ion channel subunits.
XX
XX Claim 20; SEQ ID NO 87; 347pp; English.
XX
XX This invention describes a novel method of identifying a subject
XX CC predisposed to disorder associated with ion channel dysfunction
XX CC comprising ascertaining whether at least one of the genes encoding ion
XX CC channel subunits in the subject has undergone a mutation. The invention
XX CC also describes 1) isolated nucleic acid molecules encoding an isolated
XX CC polypeptide which is a mutant or variant ion channel subunit (including a
XX CC mutant KCNQ2 subunit, where the mutation event has occurred in C terminal
XX CC domain of the subunit and leads to disturbance in the calmodulin binding
XX CC affinity of the subunit) and 2) an expression vector, cells, antibodies
XX CC and a method for producing a non-human transgenic animal which are all
XX CC used for screening of candidate pharmaceutical agents for diagnosing or
XX CC treating epilepsy or a disorder associated with ion channel dysfunction.
XX CC The mutation detected in the method disrupts the functioning of an
XX CC assembled ion channel so as to produce an epilepsy phenotype in the
XX CC subject or produces one or more disorders associated with ion channel
XX CC dysfunction such as hyper- or hypo-kalemic periodic paralysis, myotonia,
XX CC malignant hyperthermia, myasthenia, cardiac arrhythmias, episodic ataxia,
XX CC migraine, Alzheimer's disease, Parkinson's disease, schizophrenia,
XX CC anxiety, depression, phobic obsessive symptoms, neuropathic pain,
XX CC inflammatory pain, chronic/acute pain, Bartter's syndrome, polycystic
XX CC kidney disease, Dent's disease, hyperinsulinemic hypoglycemia of infancy,
XX CC cystic fibrosis, congenital stationary night blindness and total color-
XX CC blindness in the subject or to produce an epilepsy phenotype when
XX CC expressed in combination with one or more additional mutations or
XX CC variations in the ion channel subunit genes. The products of the
XX CC invention have anticonvulsant, muscular-Gen., neuroprotective,
XX CC antiarrhythmic, antidiabetic, antiparkinsonian, neuroleptic,
XX CC tranquilizer, antidepressant, analgesic, antiparkinsonian, antidiabetic and
XX CC cytoskeletal activity. This sequence represents a fragment of the human
XX CC sodium ion channel subunit SCN2A encoded by exon 20 which contains the
XX CC mutation R1319Q.
XX
XX Sequence 2005 AA;

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Query Match 60.9%; Score 6391.5; DB 9; Length 2005;
Best Local Similarity 62.1%; Pred. No. 0;
Matches 1300; Conservative 233; Mismatches 359; Indels 201; Gaps 33;

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QY 5 LLPRGTSFRFRFTRESLAAIEKMAEKOARGSTTLQESREGI,PEBEARPOLDLOASKUL 64
 Db 6 LVPBPDPDFRFTRESLAAIEORIAEBEAKRP---KQERKOBEDDENGKRPNSDLBAGSL 62
 QY 65 PDLGNPPOELIGEPELIDDPFYSTQKTFIVANKGKTI,FRSATNALVLSFPHIRAA 124
 Db 63 PFIYGDIPPEWVSFLEDDPYINKTFIIVANKKAISRFSATPALYILPFNFIRKLA 122
 QY 125 VKIIVHSLFNNMLINCTIILTNCFVMAOHDPBPWTKKVETFTAIYFESIVKILARFGCH 184
 Db 123 IKILVHSLFNNMLIKTILTNCFVFMWNSNPDMTKVETFTIYFESIKILARFGCLE 182
 QY 185 AFTPLRDPNNMLDFSVIIMAYTTEPFVLCGNVSAKRTFVRALAKTISVIGLKTIYVGLI 244
 Db 183 DTFPLRDPNNMLDFYITFAVYTEPFVLCGNVSAKRTFVRALAKTISVIGLKTIYVGLI 242
 QY 245 QGVKKLADVMVLTVECLSVFALIGLOLFNGNLRHKVR-----NFTALNGTNGSV 294
 Db 243 QGVKKLADVMVLTVECLSVFALIGLOLFNGNLRHKVR-----NFTALNGTNGSV 300
 QY 295 EADGLVME-----SIDLVSDPENYLLKNGTSDVLLCGNSSDAGTCPEGRCLKAGEHP 348
 Db 301 DENGTFNRTVSIENWDEYIEDSKSHFYFLEGGNDALLCGNSSDAGTCPEGRCLKAGEHP 360
 QY 349 DHGYTSFDSFPAFAFLFRLMTQDCWERTYQOOLRSAGKIYMI,PFMLVIFLGSFYLVNLI 408
 Db 361 NGYTSFDFPSWAFSLFRLMTQDFWENLYQOLTRAAKTYMIFFLVIFLGSFYLVNLI 420
 QY 409 LAVVANAAYEBQONATTAEIBEKKEKRFQEAEMELKKEHEALTIR-----GV 453
 Db 421 LAVVANAAYEBQONATLEBAEQKEAEFQOMLEQLKQOEBAOAAAAAASBDFSGAGSI 480
 QY 454 DTVSRSSLSMSP,LA,PNVSHS---RKRKRKRMSGGTEBEGEDRL,PKSDESDGR----- 504
 Db 481 GVFESESSVASKLSSSEKELKNRRKKKKQEQSGEEB--KNDVLKSSBEDSIRKGRFRP 539
 QY 505 -----ANMHL,SLTRGLSRTSMKPRSSRSISIFTRRR--DLGSEADPADDENS 549
 Db 540 SLEGSRLTYKRRSSPHQSLISIRGSLFSPRRSRASLPSFRGRADIGSENDPADDENS 599
 QY 550 TAGESBSHRTSLVLP--WPLRRTSAQGPSPGTSA--PGAHLGKKNSTYDCNGVSLLA 606
 Db 600 TTFDNDNSRDRDSLFPVPHGGRHSRHSNVSAQSRASRV,PI,PFMNKMSAVDCNGVSLV- 658
 QY 607 GDBEARSFGSHLRPVNLHPRPDPTTPESEBPGPQMLTQA,PCVDGEEBPGARQARLSAV 666
 Db 659 GPSTLTSAGQL-----PEGTTTETETI--RKRSSSYHVSMDLLEDFTSRQAMSTA 708
 QY 667 SVLTSALEBLEESRHKPCPCMNRLAQRYL,WECCPLWMSIKQGVKLVMNDPFTDLTITWC 726
 Db 709 SILTNMBELEBSRQKPCPCMYKPFANMCLIMDCKPKLVKHLVNLVMDPFDALITIC 768
 QY 727 IVANTLTFMALEHYNTSEFEEDMLQVGNLVTTGTF,TAEMTKI,IALDPYTFQOGNNIFDS 786
 Db 769 IVANTLTFMAHEHYMTQESSVLSVGNLVFTGIFTAMF,FKI,IAMDPYTFQOGNNIFDG 828
 QY 787 IYIVLSLMEI,GL,SM,SNL,SV,LR,SP,RL,RV,KLA,SW,TL,NL,TKI,IGNS,GAL,GN,TLV, 846
 Db 829 FIVLSLMEI,GL,AVNEGL,SV,LR,SP,RL,RV,KLA,SW,TL,NL,TKI,IGNS,GAL,GN,TLV, 888
 QY 847 AIIVFIFAVVGMOLF,GNVSE--LRDSGGL,PR,MM,MM,DF,FA,LL,IF,RL,ICGM,ETMW 904
 Db 889 AIIVFIFAVVGMOLF,GNVSE--LRDSGGL,PR,MM,MM,DF,FA,LL,IF,RL,ICGM,ETMW 948
 QY 905 DCMENVSGSLCLVFLV,VM,IGN,V,IN,LF,AL,LL,SS,PS,AD,NL,TA,PE,DE,RE,MM,NL,QL,ALA 964
 Db 949 DCMENVAGTMC,LT,FM,MM,MM,V,IG,NL,V,IN,LF,AL,LL,SS,PS,AD,NL,TA,PE,DE,RE,MM,NL,QL,ALA 1008
 QY 965 RIQGLARFVKTMTWDFCCGLLRQRPQKRAL-----AAGQ,LP,SC,IA,TP,SP,PP,ETE 1017
 Db 1009 RMQKGI,DFV,KK,IR,EF---I,KA,FR,KQ,KAL,DE,I,K,LE,LD,NK,KD,SC,IS----- 1054

QY 1018 KVPETREKTEFESE-----QPGQRP-----GDPE--PVCVPIA 1050
 Db 1055 -----NHTTIEIGKOLNYL,KD,NGT,TS,IG,GS,V,ER,KV,ND,ES,DM,YS,MM,IN,PS,LT,VY,PIA 1108
 QY 1051 VAESDTDOEBEENSLIGTEBESK,ROES,QVSGQPEAP,PD,SR,TW,SOVS,AT,AS,EA,EA,ASAS 1110
 Db 1109 VGESDPE-----NLTSEFSES,SDME-----ESKEKLNATSSSGST--- 1145
 QY 1111 QADMROQMAEP,QA,PGCGEFTPEB,CS,SGST,AD,MT,N,T,AE,LE,QL,PD,LO,GD,VK,DE,PD,CTEG 1170
 Db 1146 -----VDIGA,PA,GE,QPE-----VEPES,LE-----PEACTED 1174
 QY 1171 CVRRCPCCAVDTTQAP,GV,MM,RL,AK,TCY,HI,VS,MM,FE,FT,II,FM,LL,SS,GAL,AF,ED,LY,AE 1230
 Db 1175 CVRRFCCQ,SI,EB,SG,KL,MM,NL,RT,CKY,IV,HN,MM,FE,FT,II,FM,LL,SS,GAL,AF,ED,LY,AE 1234
 QY 1231 RKTIKVLE,VD,KM,FTYV,FV,LE,MLL,KV,VA,YG,KKY,FT,NAM,CW,LD,FL,VDV,SV,LS,VANTL 1290
 Db 1235 RKTIKVLE,VD,KM,FTYV,FV,LE,MLL,KV,VA,YG,KKY,FT,NAM,CW,LD,FL,VDV,SV,LS,VANTL 1294
 QY 1291 GFAEMGP,IK,SL,RT,RL,AL,PL,AL,SR,FG,MM,RYV,NV,NAL,GAL,PS,IM,NV,LL,VCL,IF,MS,IM 1350
 Db 1295 GYSELGA,IK,SL,RT,RL,AL,PL,AL,SR,FG,MM,RYV,NV,NAL,GAL,PS,IM,NV,LL,VCL,IF,MS,IM 1354
 QY 1351 GVN,PA,GE,GR,C,IN,OTEG,D,PL,ANTY,IV,NK,SC,ESL---NLTGEL,MYTKV,NF,PD,NY,GAG 1407
 Db 1355 GVN,PA,GE,GR,C,IN,OTEG,D,PL,ANTY,IV,NK,SC,ESL---NLTGEL,MYTKV,NF,PD,NY,GAG 1411
 QY 1408 YLALIQVATPFKGM,DM,MYA,VD,SR,GYEB,QP,WE,NLY,MY,YF,V,II,PI,IG,ST,FL,NL,PI,GV 1467
 Db 1412 YLALIQVATPFKGM,DM,MYA,VD,SR,GYEB,QP,WE,NLY,MY,YF,V,II,PI,IG,ST,FL,NL,PI,GV 1471
 QY 1468 IIDNFQOKK,CL,GG,OD,IM,TEB,QK,YY,AM,KGL,SK,KR,QK,RI,PP,P,UNK,YO,FI,DI,YTKQ 1527
 Db 1472 IIDNFQOKK,CL,GG,OD,IM,TEB,QK,YY,AM,KGL,SK,KR,QK,RI,PP,P,UNK,YO,FI,DI,YTKQ 1531
 QY 1528 AFDV,IM,FL,C,IN,MY,TM,V,VE,TDD,OS,PE,K,IN,II,AK,IN,IL,FVA,IF,GE,C,IV,KLA,AL,RH,YFT 1587
 Db 1532 VFDISIM,FL,C,IN,MY,TM,V,VE,TDD,OS,PE,K,IN,II,AK,IN,IL,FVA,IF,GE,C,IV,KLA,AL,RH,YFT 1591
 QY 1588 NSMNI,FD,RYV,NV,LS,IV,GT,LS,DI,IQ,KY,PS,PTL,FR,V,IR,LA,RI,GR,IL,IR,AK,GI,RT,LLF 1647
 Db 1592 IGMN,IF,DF,RYV,NV,LS,IV,GT,LS,DI,IQ,KY,PS,PTL,FR,V,IR,LA,RI,GR,IL,IR,AK,GI,RT,LLF 1651
 QY 1648 ALMGL,PA,LF,NIG,LL,FL,VM,FY,IS,FG,AN,PA,RYV,WE,G,ID,MM,FO,FP,AN,SM,CL,FOIT 1707
 Db 1652 ALMGL,PA,LF,NIG,LL,FL,VM,FY,IS,FG,AN,PA,RYV,WE,G,ID,MM,FO,FP,AN,SM,CL,FOIT 1711
 QY 1708 TSAGMDGL,SP,IL,NT,GP,PC,D,PTL,PN,SGS--RGDC,GS,PAV,GI,LP,FTY,II,IS,FL,IV,NMY 1766
 Db 1712 TSAGMDGL,SP,IL,NT,GP,PC,D,PTL,PN,SGS--RGDC,GS,PAV,GI,LP,FTY,II,IS,FL,IV,NMY 1771
 QY 1767 IAIILN,FS,VA,TEB,SE,TE,LE,DD,PD,MY,FI,WE,K,ED,PE,AT,O,FI,YS,V,LS,D,PA,D,AL,SE,PL,RI 1826
 Db 1772 IAIILN,FS,VA,TEB,SE,TE,LE,DD,PD,MY,FI,WE,K,ED,PE,AT,O,FI,YS,V,LS,D,PA,D,AL,SE,PL,RI 1831
 QY 1827 AKPNQ,SI,IN,MDL,PMV,SG,RI,HC,MD,IL,PA,FT,RYV,GE,SG,EM,AL,TKI,OM,EE,K,MAN,PS,KV 1886
 Db 1832 AKPNQ,SI,IN,MDL,PMV,SG,RI,HC,MD,IL,PA,FT,RYV,GE,SG,EM,AL,TKI,OM,EE,K,MAN,PS,KV 1891
 QY 1887 GYEP,IT,TL,RR,GR,ES,VA,NT,IO,AR,PH,LO,RS,LK,AS,FL,FQ,AG,SG,SE,DA,RE,GE,LT 1946
 Db 1892 GYEP,IT,TL,RR,GR,ES,VA,NT,IO,AR,PH,LO,RS,LK,AS,FL,FQ,AG,SG,SE,DA,RE,GE,LT 1949
 QY 1947 IAYV,SN,FS,RP,IG,PP,SS,IS,ST,SP,PS,YD,SV,AT,RS,DNI,LOV,RS,DY,SH,SD 1999
 Db 1950 IAD,K,AN,EN,ST-----PEK,T,MT,PS,IT,SP,PS,YD,SV,AT,RS,DNI,LOV,RS,DY,SH,SD 1995

RESULT 35
 ADY27150
 ID ADY27150 standard; protein; 2005 AA.
 XX

AC ADY27150;
 XX 05-MAY-2005 (first entry)
 XX
 DE Human SCN2A variant R223Q.
 XX
 KM SCN2A; anticonvulsant; muscular-Gen.; neuroprotective; antiarrhythmic;
 KM antimigraine; nootropic; antiparkinsonian; neuroleptic; tranquillizer;
 KM antidepressant; analgesic; nephrotoxic; antidiabetic; cytostatic;
 KM diagnostic; anxiety disorder; major depressive disorder; epilepsy;
 KM paralytic; hyperthermia; myasthenia gravis; heart arrhythmia; ataxia;
 KM migraine; Alzheimer's disease; Parkinson's disease; cystic fibrosis; pain;
 KM inflammation; polycystic kidney disease; phobia; schizophrenia;
 KM neuropathic pain; hyperglycemia; hyperinsulinemia; sodium channel;
 KM mutein.
 XX Homo sapiens.
 OS Synthetic.
 XX
 FH Key Location/Qualifiers
 FT Misc-difference 1200 /label= T1200A
 FT /note= "wild-type Thr substituted with Ala"
 XX
 PN W02005014863-A1.
 XX
 PD 17-FEB-2005.
 XX
 XX 06-AUG-2004; 2004W0-AU001051.
 XX
 PR 07-AUG-2003; 2003AU-00904154.
 XX
 XX (BION-) BIONOMICS LTD.
 XX
 PI Muller JC, Harkin LA, Dibbens LM, Phillips HA, Heron SE;
 PI Berkovic SF, Scheffer IE, Davy A;
 XX WPI: 2005-195767/20.
 DR N-PSDB; ADY7078.
 XX
 PT Identifying subject predisposed to disorder associated with ion channel
 PT dysfunction, involves determining presence of specific mutation event in
 PT genes encoding ion channel subunits.
 XX
 PS Claim 20; SEQ ID NO 66; 347pp; English.
 XX
 CC This invention describes a novel method of identifying a subject
 CC predisposed to disorder associated with ion channel dysfunction
 CC comprising ascertaining whether at least one of the genes encoding ion
 CC channel subunits in the subject has undergone a mutation. The invention
 CC also describes 1) isolated nucleic acid molecules encoding an isolated
 CC polypeptide which is a mutant or variant ion channel subunit (including a
 CC mutant KCNQ2 subunit, where the mutation event has occurred in C terminal
 CC domain of the subunit and leads to disturbance in the calmodulin binding
 CC affinity of the subunit) and 2) an expression vector, cells, antibodies
 CC and a method for producing a non-human transgenic animal which are all
 CC used for screening of candidate pharmaceutical agents for diagnosing or
 CC treating epilepsy or a disorder associated with ion channel dysfunction.
 CC The mutation detected in the method disrupts the functioning of an
 CC assembled ion channel so as to produce an epilepsy phenotype in the
 CC subject or produces one or more disorders associated with ion channel
 CC dysfunction such as hyper- or hypo-kalemic periodic paralysis, myotonias,
 CC malignant hyperthermia, myasthenia, cardiac arrhythmias, episodic ataxia,
 CC migraine, Alzheimer's disease, Parkinson's disease, schizophrenia,
 CC anxiety, depression, phobic obsessive symptoms, neuropathic pain,
 CC inflammatory pain, chronic/acute pain, Bartter's syndrome, polycystic
 CC kidney disease, Dent's disease, hyperinsulinemic hypoglycemia of infancy,
 CC cystic fibrosis, congenital stationary night blindness and total color-
 CC blindness in the subject or to produce an epilepsy phenotype when
 CC expressed in combination with one or more additional mutations or
 CC variations in the ion channel subunit genes. The products of the
 CC invention have anticonvulsant, muscular-Gen., neuroprotective,
 CC antiarrhythmic, antimigraine, nootropic, antiparkinsonian, neuroleptic,

CC tranquillizer, antidepressant, analgesic, nephrotoxic, antidiabetic and
 CC cytostatic activity. This sequence represents a fragment of the human
 CC sodium ion channel subunit SCN2A encoded by exon 19 which contains the
 CC mutation T1200A.
 XX
 XX Sequence 2005 AA;
 SQ
 Query Match 60.9%; Score 6390.5; DB 9; Length 2005;
 Best Local Similarity 62.1%; Pred. No. 0;
 Matches 1300; Conservative 232; Mismatches 360; Indels 201; Gaps 33;
 QY 5 LLEPGTSFRFRPFRSLAAIEKMAEQKQAGSTTQESBGLPEEBAAPQDLQASKKL 64
 DB 6 LVPDGPDSFRFPFRSLAAIEQIABEKAKRP---KQERDDEDENGKPNQSLDEAGKSL 62
 QY 65 PDIYGNPPOELIGEPLEDDPFTSTOKTPIVLNKGKTIIRFSATNALYLSPHPIRRA 124
 DB 63 PPIYGDIPPEMVSVPLEDDPPIYINKKTIIVLNKGAISRFSATPALYILTPNPPIRKL 122
 QY 125 VKILVHSLFNNMLIMCTILNVCVPMADHPPMVKYVEYPTATYFESLVKILARGCLH 184
 DB 123 IKILVHSLFNNMLIMCTILNVCVPMNSNPPDWTKNVEYFTGITYFESLVKILARGCLH 182
 QY 185 APTFLRDPNNWIDFSYIINAYTTEFVLDGNVSLRTERVLRALKTISVIGLKTIVGALI 244
 DB 183 DPTFLRDPNNWIDFTYITFAVTEFVLDGNVSLRTERVLRALKTISVIGLKTIVGALI 242
 QY 245 QSVKCLADVMVLTFCISVPALIGQLFMGNLRHKVR-----NFTALNGNSV 294
 DB 243 QSVKCLSDVMVLTFCISVPALIGQLFMGNLRHKCLQPPDNSFEINITSE--FNNSL 300
 QY 295 EADGLVME-----SLDLVSDPENYLKNGISDVLLCGNSSDAGCCPBGYRLKAGENP 348
 DB 301 DENGTEFNRTVSIENNDEYIEKSHFYFLEGQNDALCGNSDAGCCPBGYRLKAGENP 360
 QY 349 DGGYTSFDSFAMAFIALFRLMTQDCWERLYOQTLRSAGKIYMFPMVYFELGFSFYVNL 408
 DB 361 NQYTSFDFFSNAFLSLFRLMTQDPWENYQTLRAAGTIYMFVLVYFLGFSFYVNL 420
 QY 409 LAVVAMAYEQNATTAETBEKRFQEMEMKKEHEALTTR-----GV 453
 DB 421 LAVVAMAYEQNATTEAEQKEAEFOQMLEQKQOEBAQAAAAAASRDFSGAGGI 480
 QY 454 DTVSRSSLEMSPLAPVNSHE---RKRKRKMSSGTEGEGDRLPKSDESDGR----- 504
 DB 481 GVFSSESVASKLSSKSEKELNRRKKKKQKQSGER--KNDRLKSESDSIRKGRFR 539
 QY 505 -----AMNHLSTRLGLRTSMKPPSSRGSIPTFRRR--DLGSEADFADENS 549
 DB 540 SLEGSRLTYEKRFSSHQSLSTRGSLFSPRNSRASLFSFGRADITSENDPADERS 599
 QY 550 TAGESESHRTSLVP--WPLRTSAQGPSPTSA-PGHALHGKNSYDVCNGVSLGA 606
 DB 600 TEEDNDRSDSLFVPHRDERHSNVQSASRAVPLIPMNGKMSHSAVDCNGVSLVG- 658
 QY 607 GPPKATSPSHLRPMLNHPDITTPSEBPGQMLTSGQACVDFEPPGARQRLASV 666
 DB 659 GSTTISAQQL-----PGTTEETEI--RKRSSSYHVSMDLEPDSRQRAMSIA 708
 QY 667 SVLTSLAELEESRHKCPCKNRLAORVYIMECCPLMNSIKQGVKLWVMDPFDLITTC 726
 DB 709 SLITNMEELERSRKCPCKNRYKPNAMCLIMCCPKWLVKALVNLVMDPFDLALITTC 768
 QY 727 IVLNTLFPALAEHYNNITSEFEEMLVQGNLVFTGIFTAEMTFKIIALDPYVYFOQGNIPDS 786
 DB 769 IVLNTLFPAMEMHYPMTEGSSVLTSGNIVFTGIFTAEMFLKIIANDPYVYFOQGNIPFDG 828
 QY 787 IVLITSLMEELGSRKNSVLSRFLLVFKLAKSWPTLNTIKITGNSVGLNUTLV 846
 DB 828 FIVSLSLMEELGLANVEGSLVLSRFLLRFKLAKSWPTLNTIKITGNSVGLNUTLV 888
 QY 847 AIIIVFAVVGQQLGKQVSR--LRDSGGLPRHMDFFAFPIIFILGEMVETW 904
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Db      889 AIIIVFAVVMQRLRGSKYKCEVCCKISNDCELPRHMHDFRHSFLIVRVLGEMIEITM 948
Qy      905 DCMEEVSGSGLCLVPLLVVAVIYNLVNLFLALLSSFSADNLTAPDEDEKEMNLTQALA 964
Db      949 DCMEEVAGQMCLTVFMVAVVIGNLVYLVNLFLALLSSFSADNLTAPDEDEKEMNLTQALA 1008
Qy      965 RIQRCIRFYKRTTWDFCCGLLRQRPKPAAL-----AAQQLPSCITPFSPPPERE 1017
Db      1009 RMQKGIIDFYKRIKREF---IQAFVKRQKALDEIKPELDEJLNKKSCTIS----- 1054
Qy      1018 KVPPTKRETRFEEGE-----OPQGGTP-----GDPE-PVCVPFA 1050
Db      1055 -----NHTTIEIGKDLNLYLKQNGTTSIGSSVEKCVVDESDYMSFINNPSLTIVYVFA 1108
Qy      1051 VASDPTDQDEBDEENSLGTEEESSKQESQPVSGGPAPDSRTSGVATASSEAAASAS 1110
Db      1109 VESDPE-----NLNTEBPSSESDME-----BSKEKLTNTSSSEGST--- 1145
Qy      1111 QADWROQMAEPQAPCCGCTPEDSCSGSTADMTNTAELLBOIPDLQGVKQPEDCPTEG 1170
Db      1146 -----VDIGAPAPGEQPE-----VEPEBSIE-----PEACFTED 1174
Qy      1171 CVRRCPCCAVDTTQAPKVMWRRLKCTCYHIVESHWPETFIIFMILSSGALAPEDIYTEE 1230
Db      1175 CVAKFKCCQISTEEGKGLMMNLKACYKI VEHNMETPIVFMILSSGALAFEDYIEQ 1234
Qy      1231 RKTIKVLEAYADKFTYVFLVLEMLKMAVYGFKKYFTNACWLDPLIVDSIVSVANTL 1290
Db      1235 RKTIKTMLEYADKVFYIFILEMLKMAVAGFQVFTNACWLDPLIVDSIVSVANTL 1294
Qy      1291 GFAEMGPYKSLRLRLRLRLRALSREGRVYNVALVGPSTMTNLVCLIFMLFESM 1350
Db      1295 GVELDAIKSLRLRLRLRLRALSREGRVYNVALVGPSTMTNLVCLIFMLFESM 1354
Qy      1351 GVALPAGKPGRCINOTEGDPLNTYTYVNNKSQCESL---NLGELWYTKVAVFDVAG 1407
Db      1355 GVALPAGKPGRCINOTEGDPLNTYTYVNNKSQCESL---NLGELWYTKVAVFDVAG 1411
Qy      1408 YLALLQVATPEKGMNDIMYAAVDSRGYEEBPQWENYLYMYTYFVIFIIIPSGFPTLNFIV 1467
Db      1412 YLALLQVATPEKGMNDIMYAAVDSRGYEEBPQWENYLYMYTYFVIFIIIPSGFPTLNFIV 1471
Qy      1468 IINFNQOKKKLGGODIEMTEBOKKYNNAMKLGSKKKPKRTPRPLNKQGFEDIVTQ 1527
Db      1472 IINFNQOKKKLGGODIEMTEBOKKYNNAMKLGSKKKPKRTPRPLNKQGFEDIVTQ 1531
Qy      1528 AFDVTIMFLICLMVMTVMVETDQSPKINIILAKINLFAVFTGECIVLALRYHYEF 1587
Db      1532 VFDISTMILICLMVMTVMVETDQSPKINIILAKINLFAVFTGECIVLALRYHYEF 1591
Qy      1588 NSMNIQDFVVVILSVGTVLSDIIOKXFSPPTLFRVIRLARIGRIILIRGAGIRTLLE 1647
Db      1592 IGMNIPDFVVVILSVGTVLSDIIOKXFSPPTLFRVIRLARIGRIILIRGAGIRTLLE 1651
Qy      1648 ALMASLPALNIGLILFLVNFYISIFGMANFAYVKAAGIDDMFNQTPANSLCLPQIT 1707
Db      1652 ALMASLPALNIGLILFLVNFYISIFGMANFAYVKAAGIDDMFNQTPANSLCLPQIT 1711
Qy      1708 TSAQMDGLSPILNTGPGYCDPTLPNSNGS-RGDCGSPAYGILPFTYIIISFLIVNMV 1766
Db      1712 TSAQMDGLSPILNTGPGYCDPTLPNSNGS-RGDCGSPAYGILPFTYIIISFLIVNMV 1771
Qy      1767 IAILLENFSVATEBTEPLESDPDMFYEIMKEKPEATOFIEYSVLSDPADALSEPLRI 1826
Db      1772 IAILLENFSVATEBTEPLESDPDMFYEIMKEKPEATOFIEYSVLSDPADALSEPLRI 1831
Qy      1827 AKENQISLIMNDLPMSVSGDRICHNDILPATRYKVLGSSGEMDALKIOMEKPAAPNSKI 1886
Db      1832 AKENQISLIMNDLPMSVSGDRICHNDILPATRYKVLGSSGEMDALKIOMEKPAAPNSKI 1891
Qy      1887 SYERTITTLRKHEEVAAMVJORAFRHLIORSLSKHSFLPROQAGSGLSEEDAPREGL 1946
Db      1892 SYERTITTLRKHEEVAAMVJORAFRHLIORSLSKHSFLPROQAGSGLSEEDAPREGL 1949

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Qy      1947 IAYVSENFSPRLPSPSSSISSTSPSPSYSTRATSDNLQVRGSDYSHSED 1999
Db      1950 AIDKLTENST---PEKDTMTPTSTSPSYSTRATSDNLQVRGSDYSHSED 1995

RESULT 36
AAB99677
ID AAB99677 standard; protein, 2005 AA.
XX
AC AAB99677;
XX
DT 04-SEP-2001 (first entry)
XX
DE Human neonatal form of SCN2A protein sequence SEQ ID NO:36.
XX
KW Human; epilepsy; chromosome 2; SCN1A; SCN2A; SCN3A; identification;
KW diagnosis; mutation; chromosome 2q23-q31; neurological disorder;
KW anticonvulsant; neuroprotective.
XX
OS Homo sapiens.
XX
PN WO200138564-A2.
XX
PD 31-MAY-2001.
XX
PF 24-NOV-2000; 2000MO-CA001404.
XX
PR 26-NOV-1999; 99US-0167623P.
XX
PA (UWMC-) UNIV MCGILL.
XX
PI Rouleau GA, Lafreniere RG, Rochefort D, Cossette P, Ragsdale D;
XX
DR WPI; 2001-355945/37.
XX
DR N-PSDB; AAB55794.
XX
PT Determining a predisposition to epilepsy and/or development of epilepsy
PT comprises determining the genotype of SCN1A, SCN2A and/or SCN3A, or a DNA
PT variant, equivalent, or mutation which shows a linkage disequilibrium.
XX
PS Disclosure; Page 131-138; 268pp; English.
XX
CC The present invention describes a method (M1) of determining an
CC individual's predisposition to epilepsy and/or development of epilepsy,
CC as well as predicting the individual's response to medication. The method
CC comprises determining the genotype of at least one gene selected from
CC SCN1A, SCN2A or SCN3A, or a DNA variant, equivalent, or mutation which
CC shows a linkage disequilibrium. SCN1A, SCN2A and SCN3A are all sodium
CC channel genes located on chromosome 2. The idiopathic generalised
CC epilepsy (IGE) gene is more specifically localised on chromosome 2q23-
CC q31. Compounds identified as modulators of the biological activity of
CC SCN1A, SCN2A or SCN3A proteins or genes, are useful for treating epilepsy
CC or other neurological disorders. They have anticonvulsant and
CC neuroprotective activities. AAB55763 to AAB56164 and AAB99674 to AAB99679
CC represent SCN1A, SCN2A, and SCN3A cDNAs, gene fragments, PCR primers,
CC oligonucleotides and proteins given in the exemplification of the present
CC invention
XX
SQ Sequence 2005 AA;

Query Match 60.9%; Score 6389.5; DB 4; Length 2005;
Best Local Similarity 62.1%; Pred. No. 0;
Matches 1300; Conservative 232; Mismatches 360; Indels 201; Gaps 33;

Qy      5 LIPRGTSFRRFTRESIAIERKMAEKQARGSTTLQESRGLPBEAPRPODLQASKUL 64
Db      6 LVPFGDSFRFTRESIAIERKMAEKQARGSTTLQESRGLPBEAPRPODLQASKUL 62
Qy      65 PDLGNPQELIGPELIDPPYSTOKTFTIVANKGKTFRBSATNALYLSFPHPRRA 124
Db      63 PFIYGDIPPEWVSPLEDDPYINKKFTIVANKGKAFSFSATPALYLTPTFNPRIKLA 122

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QY 125 VKILVHSLFNNLIMCTILNLCVFNMAOHDPMPMTKYVEFTAYITFESLVIKLARGCLH 184
 DB 123 IKILVHSLFNNLIMCTILNLCVFNMAOHDPMPMTKYVEFTAYITFESLVIKLARGCLH 182
 QY 185 AFTFLRDPMMWLDPSIIMAYTTEFDLGNVSALRFRVRLAKTISVISGLKTIYVALI 244
 DB 183 DFFELRDPMMWLDPSIIMAYTTEFDLGNVSALRFRVRLAKTISVISGLKTIYVALI 242
 QY 245 QSVYKSLADVWVLYFVCSVFPALIGLQFMGNLRLKRCV-----NPLANGTNGSV 294
 DB 243 QSVYKSLADVWVLYFVCSVFPALIGLQFMGNLRLKRCV-----NPLANGTNGSV 294
 QY 295 EADGLVME-----SLDLYSDPENYLLKNGTSDVLLCGNSDPAGTPEGYCKAGENP 348
 DB 301 DNGTTRNRTVSIFFNMDEYIEDKSHFFLEBGONALICGNSSDAGCPREGIYCKAGENP 360
 QY 349 DHGYSFDSFAMAFALFRLMTODCWERLYOQTLRSAGKIYMFMLVIFLGSFYLVNLI 408
 DB 361 NGYTSFDTFSMAFLSLFRLMTODFENLYOQLRAAGKIYMFVFLVIFLGSFYLVNLI 420
 QY 409 LAVVAMAYEONQATIAETEKEKRFQAMEMLKKEHEALTIR-----GV 453
 DB 421 LAVVAMAYEONQATIAETEKEKRFQAMEMLKKEHEALTIR-----GV 453
 QY 454 DTVSRSLSEMSPLAPVNSH-----RKRKRKRMSGTEECGEDRLPKGSEDEGP----- 504
 DB 481 GVSSESSVASKLSKSEKELKRRKKKKKKKKKKKKKKKKKKKKKKKKKKKKKKKKKKKK 539
 QY 505 -----AMNHLSLTRGLSRTSMKRSSRGSIPTFRRR--DLGSEADPADDENS 549
 DB 540 SLEGSRLTYEKRFSSPHQSLSLIRGSLFSPRRNSRALFSGRAGKIGSENPADDEHS 599
 QY 550 TAEBSEBHRSLVLP--WPLRRTSAGQSPRGISA--PGHALHCKNSVUCNGVSLTGA 606
 DB 600 TFDNDSNRDLSLFPNHRGRRHSNVSQASRASVLPILPMNGKMSHAVONGVSLYG-- 658
 QY 607 GDEBATSPSGHLRLPVMLHPPTTPTEBEPGPGOMLTQAPCYDGESEBAGORALSAV 666
 DB 659 GPGTLISAGQL-----PEGTITETEI--RKRSSSYHVSMDLIEDTTSQORASIA 708
 QY 667 SVLTSALEBESRHKPCPCMNRLAORYLIMECPLMWSIKOGVKLVMMPFTDLITIMC 726
 DB 709 SILTNMBELBESRHKPCPCMYKPRANCLIMDCCKPLKVKHLVNLVMPDVLATITC 768
 QY 727 IYVNTLPMALERTNMTSEBEMQVGNLVFTGIFLTAEMTKIIALDPYTFQOGWNIIDS 786
 DB 769 IYVNTLPMALERTNMTSEBEMQVGNLVFTGIFLTAEMTKIIALDPYTFQOGWNIIDS 828
 QY 787 IYVILSIMEGLSRMSNLVLSRPSRLIRVFKLAKSWPTLNTLKIIGNSVGALGNLTIVL 846
 DB 829 FIVLSLIMEGLSRMSNLVLSRPSRLIRVFKLAKSWPTLNTLKIIGNSVGALGNLTIVL 888
 QY 847 AIIIVFPAVGMOLFCKNYSE--LRDSGSLPRMHMDPFAHFLIFRILCEMIETMW 904
 DB 889 AIIIVFPAVGMOLFCKNYSE--LRDSGSLPRMHMDPFAHFLIFRILCEMIETMW 948
 QY 905 DCMHEVSGQSLCLVFLVMTIGNLVYVNLFLALLSFSADNTLAPDEDERNNNLQIALA 964
 DB 949 DCMHEVAGQTCITLVFMMVWVIGNLVYVNLFLALLSFSADNTLAPDEDERNNNLQIALA 1008
 QY 965 RIQRGLRFVYKRTTWDFCCGLLRQRPORPAL-----AAQGLPSCIATPVSPPEETE 1017
 DB 1009 RMQKGLDFVYKRTTWDFCCGLLRQRPORPAL-----AAQGLPSCIATPVSPPEETE 1054
 QY 1018 KVPTRKETRFESE-----QPGQGP-----GDPF--PVCVPIA 1050
 DB 1055 -----NHTTIEIGKDLNLYLKDGNGTTSIGSSVKKVYVDESUYMSPINNPSLTVYVPIA 1108
 QY 1051 VASDTPDDQEBDEBNSIGTEBESSKQESQVSGGPEAPPPSRWSQVSATASSEALASAS 1110
 DB 1109 VGSSEDE-----NLTNEEFSSSESDME-----ESKEKLNATSSSEGST-- 1145
 QY 1111 QADMRQOMKAEPOAPGGGTFEPEDSCBGSFTADMTNTAELLEQIPDLGQDVKDPEDCFTEG 1170

DB 1146a-----VDIAPRABEQPE-----VEPESSE-----PEACTED 1174
 QY 1171 CVRCPCCAVDTTQAPGKQWMLRKTCTYHIVHSWETPILIMILLSSGALAFEDYLEB 1230
 DB 1175 CVRCPCCAVDTTQAPGKQWMLRKTCTYHIVHSWETPILIMILLSSGALAFEDYLEB 1234
 QY 1231 RKTITVLEADKQFVYVFLVEMLKRVVYGFCKYFPMNACMDLIDLVDSIVSLVANTL 1290
 DB 1235 RKTITVLEADKQFVYVFLVEMLKRVVYGFCKYFPMNACMDLIDLVDSIVSLVANTL 1294
 QY 1291 GFAMGPKISLRTLRALRPLRALSREGMRVNNALVGAIPSIIMNVLLVCLIFMLIFSIM 1350
 DB 1295 GYSELGAIKSLRTLRALRPLRALSREGMRVNNALVGAIPSIIMNVLLVCLIFMLIFSIM 1354
 QY 1351 GVNLPAGKRCRCINQTEBGDPLNNTYVNNKSQCESL--NLGSELWTKVKNVFNVDYAG 1407
 DB 1355 GVNLPAGKRCRCINQTEBGDPLNNTYVNNKSQCESL--NLGSELWTKVKNVFNVDYAG 1411
 QY 1408 YIALLOVAPFKGMDIMYAADVDSRGYEBQPOWEVNLVMTYFVFIIFGSEFTLNLPIGV 1467
 DB 1412 YIALLOVAPFKGMDIMYAADVDSRGYEBQPOWEVNLVMTYFVFIIFGSEFTLNLPIGV 1471
 QY 1468 IIDFNQOKKKLGGQDIPTTEBOKKYNNAMKLGSKKPKPIPRPLNKYGFIEDYVTKQ 1527
 DB 1472 IIDFNQOKKKLGGQDIPTTEBOKKYNNAMKLGSKKPKPIPRPLNKYGFIEDYVTKQ 1531
 QY 1528 APDVITMPLICLMTVMVETDQSPKINIILAKNILPVAFITGECIVKALAHVYFT 1587
 DB 1532 VFDISIMILICLMTVMVETDQSPKINIILAKNILPVAFITGECIVKALAHVYFT 1591
 QY 1588 NSWNIFDPVNVLLSYGTVLSDILGKYFSPPLFRITRLARIGRIILRGAIGITLIF 1647
 DB 1592 IGNNIFDPVNVLLSYGTVLSDILGKYFSPPLFRITRLARIGRIILRGAIGITLIF 1651
 QY 1648 ALMMSPLAFENIGLILFLVNFYISIGMANFAVYKKEAGIDMFNFOTFANSMILCFQIT 1707
 DB 1652 ALMMSPLAFENIGLILFLVNFYISIGMANFAVYKKEAGIDMFNFOTFANSMILCFQIT 1711
 QY 1708 TSAGMDGLSPILNTGPPYCDPTLPNSNGS--RDCGSPAVGILFPTTYIIISFLVVMY 1766
 DB 1712 TSAGMDGLSPILNTGPPYCDPTLPNSNGS--RDCGSPAVGILFPTTYIIISFLVVMY 1771
 QY 1767 IAILLENFSVATRESEBEPSEDDPFMYFETWEFDEAOFIYVLSDFADLSEBRLI 1826
 DB 1772 IAILLENFSVATRESEBEPSEDDPFMYFETWEFDEAOFIYVLSDFADLSEBRLI 1831
 QY 1827 AKPNOISLNMOLPMVSGDRICHOMDILFAPTRKVLGSEGMALKIOMEKFAAAPSKI 1886
 DB 1832 AKPNOISLNMOLPMVSGDRICHOMDILFAPTRKVLGSEGMALKIOMEKFAAAPSKI 1891
 QY 1887 SYEPITTLRKHEVSANVIOAFRRHLLORSLLKXASLPFROAGSGLEBDAAPREGL 1946
 DB 1892 SYEPITTLRKHEVSANVIOAFRRHLLORSLLKXASLPFROAGSGLEBDAAPREGL 1949
 QY 1947 IAYVSENSRPLGPPSSSISSTSPPSYDSTYTRATSONLQVRGSDYHSED 1999
 DB 1950 IAYVSENSRPLGPPSSSISSTSPPSYDSTYTRATSONLQVRGSDYHSED 1999
 RESULT 37
 ADY27147
 ID ADY27147 standard; protein; 2005 AA.
 AC ADY27147;
 AC XX
 DT 05-MAY-2005 (first entry)
 DT XX
 DE Human SCN2A variant R2230.
 DE XX
 KW SCN2A; anticonvulsant; muscular-Gen.; neuroprotective; antiarrhythmic;
 KW antidiabetic; antiparkinsonian; neuroleptic; tranquilizer;
 KW antidepressant; analgesic; nephrotoxic; antidiabetic; cytostatic;

diagnostic; anxiety disorder; major depressive disorder; epilepsy; paralytic; hyperthermia; myasthenia gravis; heart arrhythmia; ataxia; migraines; Alzheimer's disease; Parkinson's disease; cystic fibrosis; pain; inflammation; polycystic kidney disease; phobia; schizophrenia; neoplastic pain; hyperglycemia; hyperinsulinemia; sodium channel; mulein.

KM Homo sapiens.

OS Synthetic.

XX Key Location/Qualifiers

XX Misc-difference 223 /label= R223Q

FT /note= "wild-type Arg substituted with Gln"

PN W02005014863-A1.

PD 17-FEB-2005.

XX 06-AUG-2004; 2004WO-AU001051.

XX 07-AUG-2003; 2003AU-00904154.

PA (BION-) BIONOMICS LTD.

PI Mulley JC, Harkin LA, Dibbens LM, Phillips HA, Heron SE;

PI Berkovic SF, Scheffer IE, Davy A;

DR WPI: 2005-195767/20.

DR N-PSDB; ADY27075.

PT Identifying subject predisposed to disorder associated with ion channel dysfunction, involves determining presence of specific mutation event in genes encoding ion channel subunits.

XX Claim 20; SEQ ID NO 83; 347pp; English.

XX This invention describes a novel method of identifying a subject predisposed to disorder associated with ion channel dysfunction comprising ascertaining whether at least one of the genes encoding ion channel subunits in the subject has undergone a mutation. The invention also describes 1) isolated nucleic acid molecules encoding an isolated polypeptide which is a mutant or variant ion channel subunit (including a mutant KCNQ2 subunit, where the mutation event has occurred in C terminal domain of the subunit and 2) an expression vector, cells, antibodies and a method for producing a non-human transgenic animal which are all used for screening of candidate pharmaceutical agents for diagnosing or treating epilepsy or a disorder associated with ion channel dysfunction. The mutation detected in the method disrupts the functioning of an assembled ion channel so as to produce an epilepsy phenotype in the subject or produces one or more disorders associated with ion channel dysfunction such as hyper- or hypo-kalemic periodic paralysis, myotonia, malignant hyperthermia, myasthenia, cardiac arrhythmias, episodic ataxia, migraine, Alzheimer's disease, Parkinson's disease, schizophrenia, anxiety, depression, phobic obsessive symptoms, neuropathic pain, inflammatory pain, chronic/acute pain, Bartter's syndrome, polycystic kidney disease, Dent's disease, hyperinsulinemic hypoglycemia of infancy, cystic fibrosis, congenital stationary night blindness and total color blindness in the subject or to produce an epilepsy phenotype when expressed in combination with one or more additional mutations or variations in the ion channel subunit genes. The products of the invention have anticonvulsant, muscular-Gen., neuroprotective, anxiolytic, antidepressant, nootropic, antiparkinsonian, neuroleptic, tranquilizer, antidiabetic, analgesic, nephroprotective, antidiabetic and cytoskeletal activity. This sequence represents a fragment of the human sodium ion channel subunit SCN2A encoded by exon 6A which contains the mutation R223Q.

XX Sequence 2005 AA;

Query Match 60.9%; Score 6387.5; DB 9; Length 2005;

Best Local Similarity 62.1%; Pred. NO. 0;

Matches 1299; Conservative 233; Mismatches 360; Indels 201; Gaps 33;

QY 5 LIPGTSFRFTTEESLAIETKMAEQARGSTTLQSSREGLPREEAPRPLDQASKL 64

DB 6 LVPGPSPFRFTTESLAIETKMAEQARGSTTLQSSREGLPREEAPRPLDQASKL 62

QY 65 PDLYGNPQOELIGLEEDLDPPEYSTOKFTIYANKGTIFRESATNALVYSPHPIRAA 124

DB 63 PFIYGDIPPEAVSPLELDLPYINKKFTIYANKGAIISRSATPALYILTFPPIRKA 122

QY 125 VKIIVHSIFNNMLINCTIITNCVPAQAQDPPWTTCVETTFATITFESLVYILARGCLH 184

DB 123 IKIIVHSIFNNMLINCTIITNCVPMNSNPWTNVEVTFGTIGTIFSLIKILARGCLH 182

QY 165 APTFLRDPNNMLDSVILIMAYTTEPVDLGANVSALRTFPAVLRALKTISVISGLKTIVGALI 244

DB 163 DPTFLRDPNNMLDSVILIMAYTTEPVDLGANVSALRTFPAVLRALKTISVISGLKTIVGALI 242

QY 245 OSVKLADVNVLYTFCISVFPALIGLQIFMGILRHKCYR-----NFTALNGTNGSV 294

DB 243 OSVKLADVNVLYTFCISVFPALIGLQIFMGILRHKCYR-----NFTALNGTNGSV 300

QY 295 EADGLWE-----SLDLYSDPENYILKNGTSDVLCGNSSDAGTCPEGRCCLKAGNP 348

DB 301 DNGTTFNRTVISIFWMDVEYIEDKSHFYFLBQGNALCGNSSDAGTCPEGRCCLKAGNP 360

QY 349 DHGTSFSPFMAFPLFRMTODCERLYOQTLRSAGKIYIFPMYIFGSPFLVNL 408

DB 361 NYGTSFPTPSWAFSLFRMTODCERLYOQTLRSAGKIYIFPMYIFGSPFLVNL 420

QY 409 LAIVAMAAYEQNATIAETEKEKRFQAMENMLKKEHALTIR-----GV 453

DB 421 LAIVAMAAYEQNATIAETEKEKRFQAMENMLKKEHALTIR-----GV 480

QY 454 DTVSRSLSEMSPLAVNSHE--RSKRKRKMSSTECGDRPLPKSSSDGPR----- 504

DB 481 GVFSSESSVASKLSKSKELNRRKKKKQKQSEEB-KQDRVLKSSSESSIRKGRFP 539

QY 505 -----ANNHSLTRGLSRTSMKPPSSSGSIFTPRR--DLSGEADFPADENS 549

DB 540 SLEGRSLYERKFSPPHGLSIRGLSPRRNSASLFSRGRKDGSENDPDDHS 599

QY 550 TAGESSEHRTSLVLP--WPLRTSAQGPSPTSA-PGHALHGKNSITVDCNGVSLIGA 606

DB 600 TREDNDSRDLFLVHRHERHNSVQASRASRVPLIPMNGKMSAIDCGVSLVG- 658

QY 607 GPPEATSPGSHLRPVMLEHPDPTTPSEEPGCPQMLTSQAPCVDFEPCARORALSAV 666

DB 659 GPSTLTSAGQL-----PGTTEETEIR-RKRSSSYHVMULBDPTSRQAMSTA 708

QY 667 SVLTSLBELSESRHKPCPCNRLAQRVLIWECCEPLMMSIKQGVKLAVMDPFTDLTITWC 726

DB 709 SVLTSLBELSESRHKPCPCNRLAQRVLIWECCEPLMMSIKQGVKLAVMDPFTDLTITWC 768

QY 727 IVANTLFMALLEHYNMTSEFEEMLOVGNLVFTGIFTAEMTKIILADPYVYFOQGNIPDS 786

DB 769 IVANTLFMALLEHYNMTSEFEEMLOVGNLVFTGIFTAEMTKIILADPYVYFOQGNIPDS 828

QY 787 IVYIISLIMELGLSRMSNLVSRLRVLRYFKLAKSPNTLTIKIGNSVGLNGLTVL 846

DB 829 FIVYIISLIMELGLSRMSNLVSRLRVLRYFKLAKSPNTLTIKIGNSVGLNGLTVL 888

QY 847 AIVVFPAVNGQLPEKNYSE--LRSDSGLLPRHMDPFPAPLIIFPILGEMVETMW 904

DB 889 AIVVFPAVNGQLPEKNYSE--LRSDSGLLPRHMDPFPAPLIIFPILGEMVETMW 948

QY 905 DCMEEVGOSICLLVFLVNVYIGNLVYINLFLALLLSFSGADNLTAPDEDEMNNTQLALA 964

DB 949 DCMEEVGOSICLLVFLVNVYIGNLVYINLFLALLLSFSGADNLTAPDEDEMNNTQLALA 1008

QY 965 RIQRGLRFPVRRTTWDFCCGLLRQRPQKPAAL-----AAQGLPSCITATPSPPPETB 1017

DB 1009 RMQKIDIVFKRIREF---IQKAFVKKQALDEIKPLBDLNNKQSCIS----- 1054

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QY 1018 KVEPTRKTRFEBSGE-----QPGQGTG-----GDEP-PVCVPIA 1050
DB 1055 -----NHTIETIGKDLNLYKDGNGITSGIGSSVEKVVDESDYMSFINNPSLTVPIA 1108
QY 1051 VASDITDDQDEBENSIGTEBESKQESQVSGGPEAPDPSRTWSQVSATPASSEASAS 1110
DB 1109 VGSDEFE-----NLNTEEFSSSESDME-----ESKEKLNATSSSEGST--- 1145
QY 1111 QADWROQWKAEPQAPGCGETPEBDSCSGSGTADMTNTALTEQIPDLGQDVXDEBDCGTBG 1170
DB 1146 -----VDIGAPAGEQEP-----VEPEESIE-----DEACTED 1174
QY 1171 CVARCCCAVDTTQAPGKVMWRRLKTCYHIVESHMFEFTFIIMLLSSGALAFEDITYLER 1230
DB 1175 CVARFKCCQGISIEGKGLKMMNLRKTCYKIVENHMFETFTVPMILSSGALAFEDITYLER 1234
QY 1231 RKTIKVLEAYADKFTYFVLEMLKMWAYGPKKFTNANCMWLDPLIVDSVLSVANTL 1290
DB 1235 RKTIKTMLEAYADKFTYFVLEMLKMWAYGPKKFTNANCMWLDPLIVDSVLSVANTL 1294
QY 1291 GFAMGPIKSLRLRLRLRLRLRLRLRLRLRLRLRLRLRLRLRLRLRLRLRLRLRLRL 1350
DB 1295 GYSELDAIKSLRLRLRLRLRLRLRLRLRLRLRLRLRLRLRLRLRLRLRLRLRLRLRL 1354
QY 1351 GVNLPAGKFGRCINOTEGDLPLNTYTYNNKSOCESL---NLGELVYTKYKVPNDVAG 1407
DB 1355 GVNLPAGKFGHCINNTTGEN-FDVSVYNNYSECKALIESQOTAR---MKANVAFDVGIG 1411
QY 1408 YVALLQVATEPKGMDIMVYAAVDSRGVEBQPEWENLYMYTYFVIFIIFGSFPLNLTIGV 1467
DB 1412 YVSLLOVATEPKGMDIMVYAAVDSRVNELQPKYEDNLVYMYFVIFIIFGSFPLNLTIGV 1471
QY 1468 IIDNFQOQKKKLGCGDIFMTBEOKTYNNAMKLGSKKPKQPIRPLNKOGFTFDIVTQO 1527
DB 1472 IIDNFQOQKKKFGGGOIFMTBEOKTYNNAMKLGSKKPKQPIRPLNKOGFTFDIVTQO 1531
QY 1528 AFDVTIMPLICLMMYMTWETDQSPKINIILAKNMLFPAITGSGIYVLAALRHVYF 1587
DB 1532 VPDISIMILCLMMYMTWETDQSPKINIILAKNMLFPAITGSGIYVLAALRHVYF 1591
QY 1588 NSWNIFDFVNVILSTVGLSDIIOKYFSPFLFRVYRLRLRLRLRLRLRLRLRLRLRL 1647
DB 1592 IGMNIFDFVNVILSTVGLSDIIOKYFSPFLFRVYRLRLRLRLRLRLRLRLRLRLRL 1651
QY 1648 ALMMSLPALFNIGLILFLVMTYISIFGMANFAYVYKAGIDMNFQTPANSMLCLFQIT 1707
DB 1652 ALMMSLPALFNIGLILFLVMTYISIFGMANFAYVYKAGIDMNFQTPANSMLCLFQIT 1711
QY 1708 TSAGMGGLSPILNTGPPYCDPTLPSNNGS-RGDCGSPAVGLFTTYIIISFLVYNNY 1766
DB 1712 TSAGMGGLAPILNTPGPPCDPTLPSNNGS-RGDCGSPAVGLFTTYIIISFLVYNNY 1771
QY 1767 IAILLENFVAATEESTEPLESDDFDMFYEIWEKFEDEATQFIEVSVLSDPADLSEPLRI 1826
DB 1772 IAILLENFVAATEESTEPLESDDFDMFYEIWEKFEDEATQFIEVSVLSDPADLSEPLRI 1831
QY 1827 AAPNQISLINMDIPWYSGDRHICMDILFAFYKRVLGESGEMDALKIOMEKEMAANPSKI 1886
DB 1832 AAPNQISLINMDIPWYSGDRHICMDILFAFYKRVLGESGEMDALKIOMEKEMAANPSKI 1891
QY 1887 SYEPTITTLRRKHEEVSAMVIOQAPRRHILQSLKASFLFRQOAGSGISEBAPREBL 1946
DB 1892 STEPTITTLRRKHEEVSAMVIOQAPRRHILQSLKASFLFRQOAGSGISEBAPREBL 1949
QY 1947 IAYVSENFSPRLGPPSSSISSTSPSYDSVYTRATSDNLQVRGDSYSHSD 1999
DB 1950 LIDKLNENST----PEKTMTPSTTSPSYDSVYTRATSDNLQVRGDSYSHSD 1995

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RESULT 38
 ABB06027
 ID ABB06027 standard; protein; 2000 AA.

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XX AC ABB06027;
XX DT 10-MAY-2002 (first entry)
XX DE Human sodium channel SCN3A protein SEQ ID NO:4.
XX KM Human; sodium channel; SCN3A; chromosome 2q24-31;
XX KM familial hypercalcaemic periodic paralysis; motor endplate disease.
XX OS Homo sapiens.
XX PN WO200196552-A1.
XX PD 20-DEC-2001.
XX PF 12-JUN-2001; 2001WO-0P04956.
XX PR 13-JUN-2000; 2000JP-00177540.
XX PR 13-JUN-2000; 2000JP-00177544.
XX PA (NISC-) JAPAN SCI & TECHNOLOGY CORP.
XX PI Kanazawa I, Goto J, Jeong S;
XX DR WPI: 2002-098066/13.
XX PI N-PSDB; ABL39650.
XX PT Human sodium channels SCN1A and SCN3A and encoded genes, useful in
XX PT studying physiological mechanism in which excitant cells participate and
XX PS causes of diseases and developing drugs for motor endplate disease.
XX PS Claim 2; Page 72-81; 88pp; Japanese.
XX CC The present invention describes human sodium channels SCN1A and SCN3A.
XX CC The present sequence represents the human sodium channel SCN3A. SCN1A and
XX CC SCN3A have been located to the human chromosome 2 long arm, positions
XX CC 2q24 and 2q24-31 respectively. The sodium channel proteins are useful in
XX CC studying the physiological mechanism in which excitant cells participate
XX CC and cause human diseases, and in developing remedies for e.g. familial
XX CC hypercalcaemic periodic paralysis of extremities and motor endplate
XX CC disease
XX SQ Sequence 2000 AA;
XX
XX Query Match 60.8%; Score 6377.5; DB 5; Length 2000;
XX Best Local Similarity 62.7%; Pred. No. 0;
XX Matches 1296; Conservative 227; Mismatches 372; Indels 171; Gaps 29;
XX
XX 5 LIPRGTSSFRRTRESLIAIERKMAEKQARGSTTIOESREGPEBEAPRQDLQASKGL 64
XX 6 LVPPEPSFRLLTRESLIAIERKMAEKQARGSTTIOESREGPEBEAPRQDLQASKGL 61
XX
XX 65 PLYGNPPOELIGEPLEDDLPYVSTQKTFIVLNGKKTIFRFSATNALVYLSPHPIRRAA 124
XX 62 PLYGDIIPRMSSELEDDLPYVSTQKTFIVLNGKKTIFRFSATNALVYLSPHPIRRAA 121
XX
XX 125 VKILVHSLFNMILMTCITILNVCYMAOHDPPTKYVEYTFATLYTFESLVLKLAGFCLA 184
XX 1224KILVHSLFNMILMTCITILNVCYMAOHDPPTKYVEYTFATLYTFESLVLKLAGFCLA 181
XX
XX 185 AFTPLRDPNNMLDESVIIMATYETEVVDLGANVALTFPRYALRLKTTISVSGIKTVGALI 244
XX 182 DTFPLRDPNNMLDFSVIIMATYETEVVDLGANVALTFPRYALRLKTTISVSGIKTVGALI 241
XX
XX 245 QSVKGLADVMVLTGVCISVVALIGQLFMGNLRHKCVR-----NFTAL----- 287
XX 242 QSVKGLADVMVLTGVCISVVALIGQLFMGNLRHKCVR-----NFTAL----- 287
XX
XX 288 NGTNGSVBADGLWESLDLYLSDPENYLLKNGTSDVLLCGNSSDAGTCPEGYRCLKAGEN 347
XX 302 NGTFVNVMTSTFNMWD---YIGDSHFVYLDGOKPRLCGNSSDAGTCPEGYRCLKAGEN 358

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DR N-PSDB; ADK81761.
XX
PT New antisense compound targeted to a nucleic acid molecule encoding
PT Nav1.3, useful for useful for treating a disease or condition associated
PT with Nav1.3, e.g. pain, seizure disorder such as childhood seizure
PT disorder, or ataxia.
XX
PS Disclosure; SEQ ID NO 9096; 417bp; English.
XX
CC The present invention relates to an antisense compound targeted to a
CC nucleic acid molecule encoding Nav1.3, where the antisense compound
CC specifically hybridizes with and inhibits the expression of Nav1.3. The
CC compound and composition are useful for treating a disease or condition
CC associated with Nav1.3, e.g. pain including but not limited to
CC neuropathic pain, post-herpetic neuralgia, chronic pain, lower back pain,
CC diabetic neuropathy, trigeminal neuropathy, arthritic pain, acute pain,
CC pain from burns, migraine headache, cluster headache, mild-to-moderate
CC headache, seizure disorder such as childhood seizure disorder, including
CC but not limited to neonatal or infantile epilepsy; or ataxia. The present
CC sequence represents human Nav1.3 protein.
XX
SQ Sequence 2000 AA;
Query Match 60.8%; Score 6377.5; DB 8; Length 2000;
Best Local Similarity 62.7%; Pred. No. 0;
Matches 1296; Conservative 227; Mismatches 372; Indels 171; Gaps 29;
QY 5 LUBRGSSRRFRFRESLAAIEKMAEKQARGSTTLQESRGLPEEARPQLDLQASKL 64
DB 6 LVPGESESRFLFRESLAAIEKRAEBKAKPKKEDDN---DDEKKPKNSLLEAGKML 61
QY 65 PDLGNPPOELIGEPEDLDPEYSTQKTFVLNKGKTFPPFSATNALVYSPHPIRBA 124
DB 62 PRTYGIPEPMSEBPEEDLDPTIYINKKTFVMMKGAIRFSATSLYITPLMPVKRIA 121
QY 125 VKLVHSLFNMILMCTILTNVCMAGHDEPPMTKYVEYTFATAYTESLVKILARGFCLH 184
DB 122 IKLVHSLFNMILMCTILTNVCMAGHDEPPMTKYVEYTFATAYTESLVKILARGFCLH 181
QY 185 APTFLDPMNMDLPSYIYMAVTEPVDLGNVSLRTFRVLRALKTTISVGLKTIYCALI 244
DB 182 DEFLDPMNMDLPSYIYMAVTEPVDLGNVSLRTFRVLRALKTTISVGLKTIYCALI 241
QY 245 OSYKRLADYVVLVFCLSVFLIGLQLEPKNLRHKVR-----PFTAL----- 287
DB 242 OSYKRLADYVVLVFCLSVFLIGLQLEPKNLRHKVR-----PFTAL----- 287
QY 288 NGTNGSVEADGLVWESLDLYLSDPENYLLKNGTSDVLLCGNSSDAGTCEGYCLKAGEN 347
DB 302 NGTNGSVEADGLVWESLDLYLSDPENYLLKNGTSDVLLCGNSSDAGTCEGYCLKAGEN 358
QY 348 PHGYTSFDSFAMAFIALFLMTQDCWERYOQTLSAGKIYIMFMLVIFLGSFYLNL 407
DB 359 PHGYTSFDSFAMAFIALFLMTQDCWERYOQTLSAGKIYIMFMLVIFLGSFYLNL 418
QY 408 IIAVVMAYEBOQATIAETEEKERFOEAMENLKK-EHEALTRIGVDVYSR----- 458
DB 419 IIAVVMAYEBOQATIAETEEKERFOEAMENLKK-EHEALTRIGVDVYSR----- 458
QY 459 -----SLEMSPLAPVNSHRSKRKRMS-----SGTEBCGEDRLPKSSEDPAMNHL 509
DB 479 GELLESSSEASKLSSGAKEMNRKRQRORHLBENNGGERNSPFKSSSEDSVKRSFL 538
QY 510 SLTRGLSRTSKMP-----RSGSGITFTERR--DLGSEADPADDEN 548
DB 539 FMSDGRRLTSDKKFCPSHQLSLIRGSLFSPRNSKTSIFSFGRAKADVSEEDPADDEN 598
QY 549 STAGESSEHRTSLV--WPIRTSAQGPSPGT-SAPGHALGKNSVYDQGVVSLG 605
DB 599 STDESESRDLSLTVHRRGERNSVVSQSSMSRVPGLPANGRHSTVDQGVVSLG 658
QY 606 AGDPEATSPGSHLRVMLEHPDITTPSEBPGPOMLTSOAPCVDFEPEAGQORALSA 665

DB 659 -GPSALTSPTGOL-----PEEGTT-TETEVRKRLSSYOJSMELBEDSGORAVSI 708
QY 666AVSLTSLAELEESRHKCPCCWNRRLAQRYLIMECCPLMMSIKGVKLVWMDPTDLITM 725
DB 709 ASLTINFMELSESRKCPCCWNRRLAQRYLIMECCPLMMSIKGVKLVWMDPTDLITM 768
QY 726 CIYANTLPMALSHYNTSFEEMLOVGNLVTGSIPTAEMTFKIALDPYVYPOGNNIFD 785
DB 769 CIYANTLPMALSHYNTSFEEMLOVGNLVTGSIPTAEMTFKIALDPYVYPOGNNIFD 828
QY 786 SIIVLSIMELGSRMSNLVLRSPFLRVFKLAKSWPLTNTLKITGNSVGLNLTLY 845
DB 829 GIIVLSIMELGSRMSNLVLRSPFLRVFKLAKSWPLTNTLKITGNSVGLNLTLY 888
QY 846 LAIVTFIVVGMQLEFGKRYSE--LRDSGGLPRHMDFFAFLIRRIICGEMETM 903
DB 889 LAIVTFIVVGMQLEFGKRYSE--LRDSGGLPRHMDFFAFLIRRIICGEMETM 948
QY 904 WDMCEVSGSLCLVFLVLMVIGNLVNLFLALILSSFSADNLTPADEDEMNNDQAL 963
DB 949 WDMCEVSGSLCLVFLVLMVIGNLVNLFLALILSSFSADNLTPADEDEMNNDQAL 1008
QY 964 ARIORGRLVKKRTWDFCCGLLRQRPQAPALAAQOLPSCIATPYSPPEPEKVPTR 1023
DB 1009 GMRQKIDYVKNMRE-CFOKAFRRKPVLEIHEGKIKDSCMSNNTG-----LEISKEL 1061
QY 1024 KETRPGEPOGQPTGDE-----PVCPIVAESDITDQEBDEN 1065
DB 1062 NYLRDNGTSGVGGSSVEKXYIDENDYMFNNPSLTVTVPVIAGESDFE----- 1113
QY 1066 SLGTESESSKQESQPVSGPEAPDSRTSQAVSATASEASASQADRWQKAEPQAP 1125
DB 1114 NMTREFFSSELE-----ESKELNANSS----- 1138
QY 1126 GGGTEPESSCSGSGTAD--MTNVALELEOIPDLGODVDPEDCFTEGCVRCPCQAVDTT 1183
DB 1139 -----SEGSTVDVLPREGQAEPEPR--EDFK-PEACFTEGCIKKEPFCVSTE 1185
QY 1184 QAPGKVMRLRTCTHIYHSHWPEPTIIMILSSGALAFBDIYLEERTIKYLEYADK 1243
DB 1186 QAPGKVMRLRTCTHIYHSHWPEPTIIMILSSGALAFBDIYLEERTIKYLEYADK 1245
QY 1244 MFTYVFLVEMLEKMAVYGFKKFTNAMCMLDFLIYDVSIVSVANTLFGAEMGPISLRT 1303
DB 1246 MFTYVFLVEMLEKMAVYGFKKFTNAMCMLDFLIYDVSIVSVANTLFGAEMGPISLRT 1305
QY 1304 LRALRPLRALSRPEGRVYVVALVGAIPSIIMNVLLVCLIFMLIFSIIMGNVLPAGKRGCI 1363
DB 1306 LRALRPLRALSRPEGRVYVVALVGAIPSIIMNVLLVCLIFMLIFSIIMGNVLPAGKRGCI 1365
QY 1364 NOTEGDPLANTYITVNNKSCBSLNLTGELYWTKVKNPNNVAGYIALLOVATFKGMNDI 1423
DB 1366 NOTEGDPLANTYITVNNKSCBSLNLTGELYWTKVKNPNNVAGYIALLOVATFKGMNDI 1422
QY 1424 MYAAVDSRGYEROPOMENVLYMYIYFVPIIFSGPFTLLPFGVLIIDNPOOKKKLGOD 1483
DB 1422 MYAAVDSRGYEROPOMENVLYMYIYFVPIIFSGPFTLLPFGVLIIDNPOOKKKLGOD 1482
QY 1484 IMTEBOKKYVYAMKKLGSKKPKQKIPRELPYKQGFIPDIYTKQAFVDTIMELCLMNT 1543
DB 1483 IMTEBOKKYVYAMKKLGSKKPKQKIPRELPYKQGFIPDIYTKQAFVDTIMELCLMNT 1542
QY 1544 MMYETDDQSPKINILAKNLFLVAFPGCECLVLAALRHYPNNSMNFDPVVYVLSIV 1603
DB 1543 MMYETDDQSPKINILAKNLFLVAFPGCECLVLAALRHYPNNSMNFDPVVYVLSIV 1602
QY 1604 GTVLSDIIOKYPSPPTLFFVILARIGRLIRIRGAKGRTLLPALMMSLPALFNIGLL 1663
DB 1603 GTVLSDIIOKYPSPPTLFFVILARIGRLIRIRGAKGRTLLPALMMSLPALFNIGLL 1662
QY 1664 FLWMTYISFGMANFAYYKMEAGIDMNFQTFANSMCLFOITTSAGNDGLSLPINTG 1723
DB 1663 FLWMTYISFGMANFAYYKMEAGIDMNFQTFANSMCLFOITTSAGNDGLSLPINTG 1722

Qy	1724	PPYCDP-TLPNSNGSRDGGSPAVGILFFETYYIIISFLIVNMATIAIIILENFSVATERST	1782
Db	1723	PPDCDDPTPIHSGSSAKGDRGDPISVGIEPFVFSYIIISFLIVNMATIAVILENFSVATERSA	1782
Qy	1783	EPLESDPDMEVYELIEMKFEDEPRTAQTEIYSVLSDFADALSEPLRIAKPNQISLIIMDDLPMV	1842
Db	1783	EPLESDDEMEYEWKFEDEPRTAQTEIYSKLSDFPAALDPELLAKPNKQVLIIMDDLPMV	1842
Qy	1843	SGDRHICMDILFAPTRKVLGSGEMDALKIQMEKFKMANPSKISYEPIITTLARKKHEV	1902
Db	1843	SGDRHICMDILFAPTRKVLGSGEMDALKIQMEDEFKMANPSKISYEPIITTLARKKOREV	1902
Qy	1903	SAMVIGQAFRRHLLQRLSKHASFLEPQOAGSLSESDAPEREKGLIAYVMSNFSRPLGP	1962
Db	1903	SAATIQRFRRCLLQRLKRNISSNKKAIKG-RIDLPKODMTIIDKLGNSI---PE	1956
Qy	1963	SSSISSTSPSPSYDVTATSDNIQ	1988
Db	1957	KTDGSSSTTPPSYDVTIKPKDEKPE	1982
RESULT 40			
ADP79545	ID	ADP79545 standard; protein; 2000 AA.	
XX	AC	ADP79545;	
XX	DT	04-NOV-2004 (first entry)	
XX	DE	Human sodium III channel SCN3A splice variant.	
XX	KX	Human; sodium III channel; SCN3A; antiarrhythmic; analgesic.	
XX	OS	Homo sapiens.	
XX	PN	WO2004050857-A2.	
XX	XX	17-JUN-2004.	
XX	PD	04-DEC-2003; 2003WO-US038796.	
XX	PF	04-DEC-2002; 2002US-0431794P.	
XX	PR	(EURO-) EUROCELLIQUE SA.	
XX	PI	Kammesheidt A, Hodges D;	
XX	PI	WPI; 2004-450725/42.	
XX	DR	N-PSDB; ADP79544.	
XX	DR	GENBANK; AF225987.	
XX	PT	New human sodium III channel, useful in treating cardiac arrhythmias,	
XX	PT	herpes virus infection, diabetes mellitus, or vasculitis.	
PS	XX	Example 1; SEQ ID NO 6; 101pp; English.	
XX	XX	The present sequence is that of a previously identified splice variant of	
CC	CC	the human sodium III channel polypeptide. The sequence differs from a	
CC	CC	newly identified splice variant ADP79541 of the sodium III channel;	
CC	CC	denoted hNa1118, by 12 amino acids out of 2000. The invention provides:	
CC	CC	hNa1118 proteins and their fragments and derivatives; hNa1118-encoding	
CC	CC	nucleic acids and their fragments, including primers, probes, and	
CC	CC	hNa1118 regulatory sequences; hNa1118-specific antibodies; and methods	
CC	CC	of using these materials to detect the presence of hNa1118 proteins or	
CC	CC	nucleic acids. The invention also provides an assay method for screening	
CC	CC	to identify selective modulators of hNa1118 expression or activity.	
CC	CC	These may be useful for treatment of conditions associated with sodium	
CC	CC	channel over- or under-expression, e.g. for treatment of cardiac	
CC	CC	arrhythmia, neuronal disorders, nociceptive pain-related diseases and	
CC	CC	neuropathic pain-related diseases, e.g. pain from peripheral nerve	
CC	CC	trauma, herpes virus infection, diabetes mellitus, causalgia, plexus	
CC	CC	avulsion, neuroma, limb amputation or vasculitis.	

XX	Sequence	2000 AA	
QQ	Query Match	60.8%; Score 6377.5; DB 8; Length 2000;	
	Best Local Similarity	62.7%; Pred. No. 0;	
	Matches 1296; Conservative	227; Mismatches 372; Indels 171; Gaps 29;	
QY	5	LLPGTSFRFRPTESLSAIEKMAEAKARSTTLQESREGLPDEEAPRPOLDQAQKCL	64
DB	6	LVPEPESEFRFLPTRESLSAIEKRAAEAKAPKKEQDN---DDENKPKPSDLEAGNKL	61
QY	65	PDLXGNPQDELIGEBLEDLPDYSTOKTFIVLANKGTIFRPSATNALVYSPFHIRDA	124
DB	62	PIYVDIEPRAWSEPLDEDLYINKKTFIVANKGKAIFRPSATSALYILTPNPVRKIA	121
QY	125	VKILVHSLFPMNLINCTIITNCVPAQHPPEPWTKVETETFPATVTFESLVYILARGFCIH	184
DB	122	IKILVHSLFPMNLINCTIITNCVPMFLSNPDMTKVETETFGIYFESLKIILARGFCLE	181
QY	165	AFTPLRDPNNMLDFEVIIMAYTTEFVDIGANYSALRTPFVLPALAKTISVIGLKTIVGALI	244
DB	162	DTFLRDPNNMLDFEVIIMAYTTEFVDIGANYSALRTPFVLPALAKTISVIGLKTIVGALI	241
QY	245	ASVKKLADVMVLTVCISVFALIGLQLFPMGLRHKVR-----NFTAL-----	287
DB	242	QSVKCLSDVMILTYVCLSVFALIGLQLFPMGLRNKCLQMPEDSAFETNTTISFYNGTWD	301
QY	288	NGTNGSVEADGLWESLDLYISDPENYLLKGTSDVLLCGNSSDAGTCEGYRCLKAGEN	347
DB	302	NGTNPVNVMTSTENMMD---YIGDDSHFYVLDQXDPILCGNGSDAGQCEGYIVYAGRN	358
QY	348	PHGATSPGSAFMAFLALFRMTQDPCWELYQOULRSAGKIYMFPMVIFIGSFLYVNL	407
DB	359	PYVGTSFPTFSMAFLSLFRMTQDYEWELYULTRAQKTYMIFVLYIFIGSFLYVNL	418
QY	408	ILAVVAMAEEQNOATIAETEEKERRFOEAMEMLKK-EHEALTIRGVDTVSR-----	458
DB	419	ILAVVAMAEEQNOATLEAEQKEAFQOQMEQLKQGEAEQAVALAASASDFSGVGL	478
QY	459	-----SLEMSPLAPVNSHERRSKRRKMS---SGTECGEDRLPKPSDEGPRAMNL	509
DB	479	GELLESSSEASKLSSKGAKEMRNRRKRQREHLEBNNGERDSEPKSEDSVRSKSL	538
QY	510	SULRGLSRSMKP-----RSSRGSIFTPRRR--DLGSEADFAADEN	548
DB	539	FMSDQGRLLSDKKFCGSPHOSLSIRGSLFSPPRNSKTISFSRGRAXKQVGSNDPADDH	598
QY	549	STAGESESHRTSLVLP--WPLRRTSAQOGPSPT-SAPGHALGKNSTVDCNGVSLIG	605
DB	599	STEDDESRRDLSFVPHRGERNNSVQAQMSRMVPGLPANQKHSITVDCNGVSLVIG	658
QY	606	AGDPEATSPGSHILRFVMLEHPDPTTBBEBPGPQMLTQAFCVUGGEFEPGAROKALSA	665
DB	659	-GPSALTSLPTGO-----PREGTL-TETEVRKRLISSYQISMELEDSGKRAVSI	708
QY	666	VSTVTSALBELSESRHKCPCCWNRLLAQRVLIYECCLPMMSIKQGVKLVMNDPFTDLITM	725
DB	709	ASILTITVMELESRRQCPCCWNRFPANVFILMDCDAMLKVHLYVILVMDPFDVLATIT	768
QY	726	CIYVNLTFPMLEHYNNMTSEFEEMLYQVNLVFTGIFTAEMTKFIIALDPYVYFOQNNIFD	785
DB	769	CIYVNLTFPMAMEHYNPTGQSSVLYTGNLVFTGIFTAEMVLIKIIAMPDYVYFOQNNIFD	828
QY	786	SIIVILSLMELGLSRMSNLSVLSFRLLRVFKLAKSWPTNLTKIITIGNSVGLAGNLTIV	845
DB	829	GIIVLSLIMELGLSNVEGSLVLSFRLLRVFKLAKSWPTNLTKIITIGNSVGLAGNLTIV	888
QY	846	LAITVTFEAVNGQLRGQKQYSE--LBDSSGLPRMHMDFFPAFLIFRILGAEWIETM	903
DB	889	LAITVTFEAVVAGQLRGKSKYKECVCKINDCTLPKMHMDFFHSFLIVFRVLGAEWIETM	948
QY	904	MDCNEVSGGSLCLVFLVNVIGNLVNLFLALLSSFSADNLTAPDEDERNNITQAL	963

Db 949 WDCMEVAGOTMCLIVFMLVMVIGNLVNLPLALLLSFSSSDNLATDDNEMNNLQIAV 1008
Qy 964 ARIORGLRVRKRTTDFCCGLLRQRPQKPAALAAQOLRSCATPVSPPPTKPPR 1023
Db 1009 GMMOKGIDVKNRMB-CFOKAPFRKPKVIEIHGKNIDSCMSNNTG-----IETSKEL 1061
Qy 1024 KETREBEGROPQCGTPGDPE-----PCVPIAAESDTDQEBEEN 1065
Db 1062 NYLRONGNTTSGVGTSSVEKVIDENDYMSPIINPSLTVTVPIAVGESDPE----- 1113
Qy 1066 SLGTEBESSKQESQVPSGPEAPDPSRTWSQVATASSEAEASASQADMRQOMKAPQAP 1125
Db 1114 NLTETEFSSSESELE-----ESKEKLNATSS----- 1138
Qy 1126 GCGFTPEBDCSRGSTD--MTMTABELLEQIPDLGQVYKDPBDCFTGCVRGCPCAVDTT 1183
Db 1139 -----SEGSTVDVVLPRREGQAEPEP--EDFK--PEACFTEGCIKKPPCQVSTE 1185
Qy 1184 QAPGVWMLRKTCTYHIVHSWPEFPIIFMILSSGALAFEDIYLEERTIKVLEAYDK 1243
Db 1186 BKGKIMWMLRKTCTYHIVHSWPEFPIIFMILSSGALAFEDIYLEERTIKVLEAYDK 1245
Qy 1244 MFTYFVLEMLKMWAYGFKYFTNAWCWLDPLIVDSLSVANTLGFAPKGPISKLT 1303
Db 1246 VFTYFVLEMLKMWAYGFKYFTNAWCWLDPLIVDSLSVANTLGFAPKGPISKLT 1305
Qy 1304 LRALPLRALSREBGRVNVNLCALPISIMNVLVCLIFMLIFSIMGNLPAKFKRCI 1363
Db 1306 LRALPLRALSREBGRVNVNLCALPISIMNVLVCLIFMLIFSIMGNLPAKFKRHCV 1365
Qy 1364 NOTEGDLPLANTYIVNNKSOCESLNTGELYTKVKVNFDPNVGAGYLALLQVATPKGMDI 1423
Db 1366 NMTTGMM-FDISDVNNLSDCQALG--KQARMQNVKNFDPNVGAGYLALLQVATPKGMDI 1422
Qy 1424 MYAAVDSRGYEQPQWEXNLWYIYFVIFIFGSEFTLNLFIGVIIDNFNOQKKLGQGD 1483
Db 1423 MYAAVDSRDLKQPYVBENLYWYLFVIFIFGSEFTLNLFIGVIIDNFNOQKKLGQGD 1482
Qy 1484 IFMTBEOKKYNYAMKLGSKKQKPIPRANKQOQWDFVTRQVFDISIMILICLWYT 1543
Db 1483 IFMTBEOKKYNYAMKLGSKKQKPIPRANKQOQWDFVTRQVFDISIMILICLWYT 1542
Qy 1544 MMVETDQSPKIKINILFVAIFTEGCIKLAALRHYFTNSMNI FDPVVVILSTV 1603
Db 1543 MMVETDQSKMTLVLSKINLVFVIFTEGCEYKLSLRHYFTTGMNIFDFVVILSTV 1602
Qy 1604 GTVLSDIIOKYFPSPTLFRVIRLARIGRILRLIRGAKGIRTLFPALMMSLPALFNIGLL 1663
Db 1603 GMLFLAEMIEKYSVSTLFRVIRLARIGRILRLIRGAKGIRTLFPALMMSLPALFNIGLL 1662
Qy 1664 FLVMTIYISIFGMANPAYKMEAGIDMNFOTFANSMLCFQITTSAGMDGLSPILANTG 1723
Db 1663 FLVMTIYAIFGMSNPAKYKKEGIDMNFETFGNSMICLFOITTSAGMDGLAPILNSA 1722
Qy 1724 PPYCDP-TLPNSNGSRGCGSPAVGILFTTYIIISPLIVVMYIAIILENSVATERST 1782
Db 1723 PPDCDPDTIHPGSSVYKGRDBSVGIFFFVSTIISFLVVMYIAIILENSVATERESA 1782
Qy 1783 EPLSEDDPDMYEIWEKPEATOFIBYSVLSDFADALSEPLRIAKPNOISLINDLPMV 1842
Db 1783 EPLSEDDPEMFYEVWEKFDPAOTGTEBFPSKLSDFADALDPLLIAPKPVQIAMDLPV 1842
Qy 1843 SGRDRHCHMDILPAFTKRVLGSGEMDALKIOMEKFPMAANPSKISYEPITTTLRKHEEV 1902
Db 1843 SGRDRHCHMDILPAFTKRVLGSGEMDALRIQMEDRFMAANSKYSYEPITTTLRKHEEV 1902
Qy 1903 SAMVIOBAFRBHLORSLKSHASFLRQOAGSLSEEDA PERBGLIAYVMSNFSPRLGP 1962
Db 1903 SAAIQRNFRCTYLLKORLKNISSNYKKAIRK--RIDLPKODMIIIDKLANGST---PE 1956
Qy 1963 SSSSISSTSPPSYDSTRTATSDNLQ 1988
Db 1957 KTDGSSSTPPPSYDSTTKPKPEKEPE 1982

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